

☐ 3. Document ID: US 6379896 B1

L1: Entry 3 of 20

File: USPT

Apr 30, 2002

US-PAT-NO: 6379896

DOCUMENT-IDENTIFIER: US 6379896 B1

TITLE: Probes for variance detection

Full	Title	Citation	Front	Review	Classification	Date	Reference	Sequences	Attachments	Claims	KMCD
Draw Desc	Image										

☐ 4. Document ID: US 6307039 B1

L1: Entry 4 of 20

File: USPT

Oct 23, 2001

US-PAT-NO: 6307039

DOCUMENT-IDENTIFIER: US 6307039 B1

TITLE: Method for analyzing a polynucleotide containing a variable sequence and a set or array of oligonucleotides therefor

Full	Title	Citation	Front	Review	Classification	Date	Reference	Sequences	Attachments		KMCD
Draw Desc	Image										

☐ 5. Document ID: US 6268142 B1

L1: Entry 5 of 20

File: USPT

Jul 31, 2001

US-PAT-NO: 6268142

DOCUMENT-IDENTIFIER: US 6268142 B1

TITLE: Diagnostics and therapeutics for diseases associated with an IL-1 inflammatory haplotype

Full	Title	Citation	Front	Review	Classification	Date	Reference	Sequences	Attachments		KMCD
Draw Desc	Image										

☐ 6. Document ID: US 6238863 B1

L1: Entry 6 of 20

File: USPT

May 29, 2001

US-PAT-NO: 6238863

DOCUMENT-IDENTIFIER: US 6238863 B1

TITLE: Materials and methods for indentifying and analyzing intermediate tandem repeat DNA markers

Full	Title	Citation	Front	Review	Classification	Date	Reference	Sequences	Attachments		KMCD
Draw Desc	Image										

☐ 7. Document ID: US 6183958 B1

L1: Entry 7 of 20

File: USPT

Feb 6, 2001

US-PAT-NO: 6183958

DOCUMENT-IDENTIFIER: US 6183958 B1

TITLE: Probes for variance detection

Full	Title	Citation	Front	Review	Classification	Date	Reference	Sequences	Attachments
Draw Desc	Image								

KIMC

☐ 8. Document ID: US 6165716 A

L1: Entry 8 of 20

File: USPT

Dec 26, 2000

US-PAT-NO: 6165716

DOCUMENT-IDENTIFIER: US 6165716 A

TITLE: Screening for disorders of serotonergic dysfunction

Full	Title	Citation	Front	Review	Classification	Date	Reference	Sequences	Attachments
Draw Desc	Image								

KIMC

☐ 9. Document ID: US 6150095 A

L1: Entry 9 of 20

File: USPT

Nov 21, 2000

US-PAT-NO: 6150095

DOCUMENT-IDENTIFIER: US 6150095 A

TITLE: Method for analyzing a polynucleotide containing a variable sequence

Full	Title	Citation	Front	Review	Classification	Date	Reference	Sequences	Attachments
Draw Desc	Image								

KIMC

☐ 10. Document ID: US 6132724 A

L1: Entry 10 of 20

File: USPT

Oct 17, 2000

US-PAT-NO: 6132724

DOCUMENT-IDENTIFIER: US 6132724 A

TITLE: Allelic polygene diagnosis of reward deficiency syndrome and treatment

Full	Title	Citation	Front	Review	Classification	Date	Reference	Sequences	Attachments
Draw Desc	Image								

KIMC

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Term	Documents
VNTR.DWPI,EPAB,JPAB,USPT,PGPB.	306
VNTRS.DWPI,EPAB,JPAB,USPT,PGPB.	135
ALLELE.DWPI,EPAB,JPAB,USPT,PGPB.	7781
ALLELES.DWPI,EPAB,JPAB,USPT,PGPB.	7461
PCR.DWPI,EPAB,JPAB,USPT,PGPB.	32842
PCRS.DWPI,EPAB,JPAB,USPT,PGPB.	942
(VNTR SAME ALLELE SAME PCR).USPT,PGPB,JPAB,EPAB,DWPI.	20
(VNTR SAME ALLELE SAME PCR).USPT,PGPB,JPAB,EPAB,DWPI.	20

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UC had an effect on the risk of developing pouchitis. PATIENTS: We determined the genotypes of the IL-1RN and HLA DR beta and DQ beta loci for 28 subjects with previous UC and a pouch with no evidence of pouchitis for a minimum of 2 years after formation of an ileo-anal reservoir (mean 6.3 years; range 2-17 years) and 25 subjects with previous UC and pouchitis confirmed by strict histological examination of pouch mucosal biopsy. The IL-1RN genotypes were also determined for 86 healthy controls and 61 unrelated patients with familial adenomatous polyposis (FAP). The p-ANCA status was determined for all 25 pouchitis subjects but only 23/28 non-pouchitis subjects, with 15 unaffected subjects as a negative control. METHODS: The HLA haplotypes of the UC groups were determined by polymerase chain reaction sequence-specific primer (PCR-SSP) typing and the IL-1RN genotypes were determined by PCR and agarose gel electrophoresis. The p-ANCA status was determined by indirect immunofluorescence. RESULTS: A chi 2 of 5.686 with 1 degree of freedom and a P value of 0.0171 using Yates' correction was obtained by comparing the IL-1RN allele frequencies of the combined UC groups to the FAP controls, and a chi 2 of 6.801 with 1 degree of freedom and a P value of 0.0091 comparing the pouchitis group to the FAP controls. The HLA haplotype frequencies did not vary significantly between groups nor did they correlate with p-ANCA status. There were also no significant associations of the p-ANCA status and pouchitis. CONCLUSION: There is an increased frequency of IL-1RN allele 2 in UC, with the majority of the association arising from the pouchitis group, suggesting that the presence of allele 2 in patients with UC affects the disease outcome. However, the HLA frequencies and p-ANCA status do not have any significant associations.

5/3,AB/19 (Item 19 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

09090828 96387392 PMID: 8794927

Genetic typing by capillary electrophoresis with the allelic ladder as an absolute standard.

Zhang N; Yeung ES

Ames Laboratory-USDOE and Chemistry Department, Iowa State University, Ames 50011, USA.

Analytical chemistry (UNITED STATES) Sep 1 1996, 68 (17)

p2927-31, ISSN 0003-2700 Journal Code: 4NR

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

We demonstrate a genetic typing method based on capillary electrophoresis/laser-induced fluorescence (CE-LIF). VNTR polymorphism in the human D1S80 locus was studied. A pooled allelic ladder, which contains the 27 most common human alleles, was used as the absolute standard. Extracted genomic DNA from an individual was amplified by polymerase chain reaction (PCR). Typing can be accomplished by co-injection of the PCR product and the D1S80 ladder and then running CE. Separation by a polymer solution of poly(ethylene oxide) in uncoated fused-silica capillaries allows high-resolution, repeated runs in the same capillary. Sensitive detection with minimal sample preparation is possible by using ethidium bromide as the intercalating dye. Statistical analysis of the data indicates a high level of confidence in matching the bands despite variations in the injection process or in the CE system. Future adaptation to a multiple-capillary array system should allow high-speed, high-throughput operation.

5/3,AB/20 (Item 20 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

09047983 96406629 PMID: 8810744

The **allele** frequency distribution at **VNTR** locus D17S5 (YNZ22) in a Japanese population sample.
Harashima N; Ota M; Katsuyama Y; Sugiyama E; Liu C; Fukushima H
Department of Legal Medicine, Shinshu University, School of Medicine, Matsumoto, Japan.

Nippon hoigaku zasshi (JAPAN) Aug 1996, 50 (4) p237-40, ISSN 0047-1887 Journal Code: KL3

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

The **allele** frequencies for the YNZ22 locus were determined in a Japanese population sample (n = 164) using the polymerase chain reaction (PCR). We observed 10 **alleles** and 39 genotypes. **Allele** distributions in our data showed a different pattern from those reported in the literature for European Caucasians. No deviation from Hardy-Weinberg equilibrium was found. The observed heterozygosity was 83.5%. In Japan the YNZ22 is one of useful genetic markers for paternity testing and identify testing, with a chance of exclusion (CE) value of 68% and a power of discrimination (PD) value of 89%.

5/3,AB/21 (Item 21 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

09047977 96304139 PMID: 8752989

Long-distance PCR of **VNTR** at the D17S74 (CMM86) locus.

Kishida T; Tamaki Y; Kuroki K

Department of Forensic Medicine, Oita Medical University, Japan.

Nippon hoigaku zasshi (JAPAN) Jun 1996, 50 (3) p174-7, ISSN 0047-1887 Journal Code: KL3

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

We have successfully amplified D17S74 (CMM86) **alleles** by a long-distance polymerase chain reaction (PCR) using TaKaRa Ex Taq (a Taq DNA polymerase with a 3'-exonuclease activity) and Perfect Match Polymerase Enhancer (a special polymerase enhancer). We adopted a hot-start technique with TaqStart antibody. Because of the high guanine content (60%) in D17S74 **alleles**, removal of K⁺ from the buffers was quite effective. The use of K(+)-free buffers reduces premature chain termination in G-rich regions, thereby facilitating amplification of targets containing such sequences. The 17 **alleles** amplified from DNA samples of 72 unrelated Japanese subjects ranged from 1.05 to 3.5 kb, with a heterozygosity of 92%. PCR amplification of D17S74 **alleles** makes their detection simpler than by conventional Southern blotting, and increases the practical utility of the locus.

5/3,AB/22 (Item 22 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

09002738 96438637 PMID: 8840997

Internal sequence variations in the Ha-ras variable number tandem repeat rare and common **alleles** identified by minisatellite variant repeat polymerase chain reaction.

Conway K; Edmiston SN; Hulka BS; Garrett PA; Liu ET

Department of Epidemiology, The University of North Carolina at Chapel Hill, 27500, USA.

Cancer research (UNITED STATES) Oct 15 1996, 56 (20) p4773-7, ISSN 0008-5472 Journal Code: CNF

Contract/Grant No.: P50 CA58223, CA, NCI

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

In this report, we describe the sequence allelotyping of the Ha-ras variable number tandem repeat (VNTR) region using a minisatellite variant repeat (MVR)-PCR approach. This method permits the rapid identification of internal sequence variations among the VNTR alleles, exploiting the presence of two polymorphic sites within the 28-bp repeat subunits that give rise to four distinct repeat types. Using MVR-PCR, 20 to 25 repeats at the 5' end of the VNTR can be sequenced rapidly and reliably. MVR typing of the common alleles a1, a2, a3, and a4 shows that the first six repeats at the 5' end of each allele constitutes an invariant region. Beginning with repeat 7, characteristic "signature" MVR patterns emerge for each common allele. The a1 and a2 common alleles were found to consist of specific repeat types 1, 2, and 3, whereas a3 and a4 contain an additional repeat type 4 not present in the smaller alleles. MVR typing of rare-length alleles indicates that they are comprised of disorganized sequences, although they usually bear a resemblance to one of the common alleles at the 5'-most end. These results suggest that the rare alleles may be generated from recombination or gene conversion-type events involving the common progenitor alleles. MVR typing could, therefore, improve the ascertainment of rare Ha-ras alleles and may provide molecular insights into the genesis of cancer-associated alleles.

5/3,AB/23 (Item 23 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

08984112 96413842 PMID: 8816977

Imprinted and genotype-specific expression of genes at the IDDM2 locus in pancreas and leucocytes.

Vafiadis P; Bennett ST; Colle E; Grabs R; Goodyer CG; Polychronakos C
Department of Pediatrics, McGill University, Montreal, Quebec, Canada.
Journal of autoimmunity (ENGLAND) Jun 1996, 9 (3) p397-403,
ISSN 0896-8411 Journal Code: ADL

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

One of the loci encoding susceptibility to insulin-dependent diabetes mellitus (IDDM) is IDDM2, mapped to a variable number of tandem repeats (VNTR) polymorphism situated 596 bp upstream of the insulin gene (INS). The shorter alleles (class I) predispose to IDDM, while the longer class III alleles are protective. Besides INS, it is possible that transcription levels of IGF2, the nearby gene encoding the insulin-like growth factor II, may be modulated by allelic forms of the VNTR. In an effort to define the pathophysiologic mechanism of the IDDM2 effect, we examined the effect, in cis, of VNTR genotype on steady-state mRNA levels of INS in samples of human fetal pancreas, and of IGF2 in leucocytes of diabetic children. Relative levels of mRNA transcripts derived from each chromosome carrying a defined VNTR allele were measured by RT-PCR, taking advantage of transcribed polymorphisms at the 3' untranslated region of each gene. In 10 samples of human fetal pancreas, INS transcripts from chromosomes carrying a class III VNTR were slightly but significantly ($P = 0.015$) lower than those from class I (13% lower, 95% confidence limits 3-21%). In 10 leucocyte samples, mRNA from both IGF2 alleles was seen, indicating relaxation of the parental imprinting of IGF2 in these cells. However, this relaxation was incomplete as maternal allele mRNA was systematically at a lower level than paternal. The paternal/maternal ratio varied widely among individual subjects. Two of the most extreme cases, demonstrating almost complete repression of the maternal allele, were identical twins, suggesting that this variable relaxation of imprinting is genotype-dependent. However, this genotype-dependence cannot be accounted for by the maternal VNTR, as the mean ratios of paternal/maternal IGF2 mRNA levels were not statistically different in individuals with a maternal VNTR of class I vs. class III (3.2 ± 1.5 vs. $3.89 \pm$

0.94). Thus, we present evidence that: (a) class III **VNTR** **alleles** are associated with lower INS mRNA in fetal pancreas than class I **alleles**. The biologic importance of this difference remains to be determined; and (b) the variable relaxation of IGF2 imprinting seen in human leucocytes is not dependent on the presence of a class I vs. a class III **VNTR**.

5/3,AB/24 (Item 24 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

08975664 96285882 PMID: 8721432

High-resolution vertical PAGE: an alternative electrophoretic system with multiple forensic applications.

Schneider HR; Rand S

Hessisches Landeskriminalamt, Wiesbaden, Germany.

International journal of legal medicine (GERMANY) 1996, 108 (5)

p276-9, ISSN 0937-9827 Journal Code: AX1

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

DNA profiling based on **PCR** technology has become a powerful tool in forensic casework, as it enables specific amplification of polymorphic **VNTR** loci from very small quantities or even degraded human DNA. Genetic typing of AmpFLP or STR loci may require electrophoretic separation techniques usually achieved by sequencing gels. In this report we present a non-denaturing high-resolution vertical PAGE system which can be easily used for both **VNTR** subgroups. The system has been evaluated for single- and multiplex use in routine casework and has been shown to be rapid, sensitive and reproducible.

5/3,AB/25 (Item 25 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

08975626 96335705 PMID: 8755922

Mutation rate in the hypervariable **VNTR** g3 (D7S22) is affected by **allele** length and a flanking DNA sequence polymorphism near the repeat array.

Andreassen R; Egeland T; Olaisen B

Institute of Forensic Medicine, University of Oslo, Norway.

American journal of human genetics (UNITED STATES) Aug 1996, 59

(2) p360-7, ISSN 0002-9297 Journal Code: 3IM

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

The hypervariable human minisatellite locus D7S22 (g3) is highly polymorphic. The allelic distribution in D7S22 features a size clustering of the **alleles** and a comparably low allelic diversity among small **alleles**. This reduced diversity could reflect a situation where some **alleles** are less likely to mutate than others. Several factors could explain such an effect, including **allele** size, variation in repeat composition, and allelic differences in nearby cis-acting elements affecting the mutation rate. We have characterized 40 de novo mutations found on Southern blots in a large amount of paternity-testing material. There is a significant excess of paternal mutations, and small size changes are most frequent. Mutation rate is affected by **allele** length, with highest rates in larger **alleles**. **Alleles** of the family groups with D7S22 mutations and 50 small **alleles** were analyzed by nucleotide sequencing. Two hundred thirty-six base pairs of the immediate flanking region upstream of the repeat array were **PCR** amplified and screened for point mutations by DNA sequencing of the **PCR** products. Two base substitution polymorphisms were identified: one C/G transversion and one A/G transition, 54 bp and 173 bp upstream of the repeat array,

respectively. There is a significant association between mutation and occurrence of 54C, while association is not obvious between mutation rate and the 173A/G variants. There is a marked association between different flanking haplotypes and allele size, and within the smallest allele-size group, all alleles had the 54G/173A haplotype. Both allele size and allelic state at site 54 remain associated with mutation rate when the other factor is controlled. Possible mechanisms behind the variation in mutation rate in D7S22 are discussed.

5/3,AB/26 (Item 26 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

08975057 96328303 PMID: 8707305

Identification of a variable number tandem repeat region in the human T cell receptor alpha-delta (TCRAD) locus.

Buchmayer H; Rumpold H; Mannhalter C

Department of Laboratory Medicine, General Hospital of Vienna, University of Vienna, Austria.

Human genetics (GERMANY) Sep 1996, 98 (3) p333-5, ISSN 0340-6717 Journal Code: GED

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

A number of different polymorphisms have been observed in coding as well as in non-coding regions of T cell receptor (TCR) genes. We report the identification and characterization of a highly polymorphic locus in the 3' noncoding region of the human T cell receptor alpha/delta (TCRAD) on chromosome 14. In 202 unrelated individuals, ten different alleles

were distinguished by polymerase chain reaction (PCR) and a heterozygosity rate of 64% was calculated. Sequence analysis revealed that this polymorphic region consists of 10 bp imperfect repeat units and represents a variable number tandem repeat region (VNTR). Stable Mendelian inheritance of this novel polymorphic marker was proven in four families. The localization of this VNTR polymorphism in the TCRAD locus should make it a useful system for linkage analysis in immunological disorders with a known role of TCRAD.

5/3,AB/27 (Item 27 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

08973899 96251121 PMID: 8665685

Apolipoprotein B 3' hypervariable repeat genotype: association with plasma lipid concentration, coronary artery disease, and other restriction fragment polymorphisms.

Wu JH; Chern MS; Lo SK; Wen MS; Kao JT

Molecular Genetic Laboratory 3, Chang Gung Medical College, Taiwan. jhwu@cguapo.cgu.edu.tw

Clinical chemistry (UNITED STATES) Jun 1996, 42 (6 Pt 1) p927-32, ISSN 0009-9147 Journal Code: DBZ

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

Apolipoprotein B gene 3' variable number tandem repeat (VNTR) and related regions were amplified by PCR and analyzed by agarose gel electrophoresis. Eighteen VNTR alleles (VNTR25, 26, 29, 31, 33, 35, 37, 39, 41, 43, 45, 47, 49, 51, 53, 55, 58, 60) and 45 genotypes were observed in 477 Taiwanese subjects. The VNTR35 allele and genotype VNTR35/35 were observed most frequently in this population. The polymorphism information content was 0.62. Some minor alleles, such as VNTR25 and 60, were found only in coronary artery disease (CAD) and stroke patients in our sampling, and no statistically significant difference was observed in VNTR allelic frequency between control and

CAD or stroke patients. Significant differences in allelic distribution of some VNTR alleles were observed between our normal Taiwanese population and a Caucasian group studied by others. VNTR43-47 and AluI+ (coding Ala591) restriction fragment length polymorphism (RFLP) as well as VNTR49-60 and EcoRI- (coding Lys4154) RFLP were found to be highly coinherited. No apparent association between the VNTR genotype and plasma lipid concentration was observed; however, for the same genotype, the CAD and stroke patients frequently showed higher lipoprotein(a) and lower HDL cholesterol concentrations than the control group.

5/3,AB/28 (Item 28 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

08973650 96239826 PMID: 8652419

Estimating minimum allele frequencies for DNA profile frequency estimates for PCR-based loci.

Budowle B; Monson KL; Chakraborty R
Forensic Science Research and Training Center, FBI Laboratory, FBI Academy, Quantico, Virginia 22135, USA.

International journal of legal medicine (GERMANY) 1996, 108 (4)
p173-6, ISSN 0937-9827 Journal Code: AX1

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

In order that there can be confidence that DNA profile frequency estimates will not place undue bias against a defendant, 2 methods are described for estimating minimum allele frequency bounds for PCR-based loci. One approach estimates minimum allele frequencies for VNTR and STR loci using sample size and the observed heterozygosity at a locus, while the second approach, appropriate for loci typed with allele-specific oligonucleotide probes, is based only on sample size. The use of a minimum allele frequency enables compensation for sparse sampling of infrequent alleles in population databases.

5/3,AB/29 (Item 29 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

08961839 96288945 PMID: 8754581

Paternity exclusion by DNA markers: effects of paternal mutations.

Chakraborty R; Stivers DN

Human Genetics Center, School of Public Health, University of Texas-Houston Health Science Center, USA.

Journal of forensic sciences (UNITED STATES) Jul 1996, 41 (4)
p671-7, ISSN 0022-1198 Journal Code: I5Z

Contract/Grant No.: GM 41399, GM, NIGMS; GM 45861, GM, NIGMS

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

In parentage testing when one parent is excluded, the distribution of the number of loci showing exclusion due to mutations of the transmitting alleles is derived, and it is contrasted with the expected distribution when the exclusion is caused by nonpaternity. This theory is applied to allele frequency data on short tandem repeat loci scored by PCR analysis, and VNTR data scored by Southern blot RFLP analysis that are commonly used in paternity analysis. For such hypervariable loci, wrongly accused males should generally be excluded based two or more loci, while a true father is unlikely to be excluded based on multiple loci due to mutations of paternal alleles. Thus, when these DNA markers are used for parentage analysis, the decision to infer non-paternity based on exclusions at two or more loci has a statistical support. Our approach places a reduced weight on the combined

exclusion probability. Even with this reduced power of exclusion, the probability of exclusion based on combined tests on STR and VNTR loci is sufficiently large to resolve most paternity dispute cases in general populations.

5/3,AB/30 (Item 30 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

08890124 95207158 PMID: 7899279

A simple, rapid, and highly informative PCR-based procedure for prenatal diagnosis and carrier screening of phenylketonuria.

Eisensmith RC; Goltsov AA; Woo SL

Department of Cell Biology, Baylor College of Medicine, Houston, TX 77030.

Prenatal diagnosis (ENGLAND) Dec 1994, 14 (12) p1113-8, ISSN

0197-3851 Journal Code: PJ7

Contract/Grant No.: HD-17711, HD, NICHD

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

The polymorphic information content (PIC) and the degree of heterozygosity of several polymorphic systems within the phenylalanine hydroxylase (PAH) gene were determined in 85 European Caucasian and 19 Chinese phenylketonuria (PKU) kindreds. The first system examined, a short tandem repeat (STR), had a PIC of 80 and 73 per cent in these Caucasian and Chinese samples, respectively. The degree of heterozygosity actually observed for this system was 81 and 64 per cent in the Caucasian and Chinese PKU families, respectively. Through the addition of a second polymorphism based on a variable number of tandem repeats (VNTR), the PIC was increased to 90 per cent in Caucasians, but only to 75 per cent in Chinese. The degree of heterozygosity observed for this combination was 94 per cent in European PKU families and 67 per cent in Chinese PKU families. The further addition of an XmnI RFLP increased both the PIC and the level of heterozygosity in Caucasians to 95 per cent, but did not change either of these measures in Chinese. The combined use of these three polymorphisms significantly increases the informativity of prenatal diagnostic and carrier screening procedures in both Caucasian and Chinese PKU kindreds. Furthermore, since each of these polymorphisms can be studied by PCR-based methods, these new tests can be performed more quickly and easily than previous Southern-based procedures.

5/3,AB/31 (Item 31 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

08890123 95207148 PMID: 7899270

Prenatal diagnosis by minisatellite analysis in Italian families with phenylketonuria.

Romano V; Dianzani I; Ponzzone A; Zammarchi E; Eisensmith R; Ceratto N; Bosco P; Indelicato A

Istituto OASI (I.R.C.C.S.), Troina, Italy.

Prenatal diagnosis (ENGLAND) Oct 1994, 14 (10) p959-62, ISSN

0197-3851 Journal Code: PJ7

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

A polymorphic short tandem repeat (STR) in intron 3 (Goltsov et al., 1993) and a variable number of tandem repeats (Hind III-VNTR) flanked by two constant Hind III sites (Golstov et al., 1992) have been recently identified in the human phenylalanine hydroxylase (PAH) gene. These polymorphisms are easily detected by the polymerase chain reaction (PCR) and gel electrophoresis. We report on the use of these two novel polymorphisms in three Italian families with pregnancies at risk for

classical phenylketonuria (PKU). A carrier status for PKU was ascertained in two fetuses; the third family refused prenatal diagnosis, although informativeness was shown to be complete.

5/3,AB/32 (Item 32 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

08881326 96216146 PMID: 8651279

Mucopolysaccharidosis IVA: four new exonic mutations in patients with N-acetylgalactosamine-6-sulfate sulfatase deficiency.

Tomatsu S; Fukuda S; Yamagishi A; Cooper A; Wraith JF; Hori T; Kato Z; Yamada N; Isogai K; Sukegawa K; Kondo N; Suzuki Y; Shimozawa N; Orii T

Department of Pediatrics, Gifu University School of Medicine, Gifu, Japan.

American journal of human genetics (UNITED STATES) May 1996, 58

(5) p950-62, ISSN 0002-9297 Journal Code: 3IM

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

We report four new mutations in Japanese patients with mucopolysaccharidosis IVA (MPSIVA) who were heterozygous for a common double gene deletion. A nonsense mutation of CAG to TAG at codon 148 in exon 4 was identified, resulting in a change of Q to a stop codon and three missense mutations. V (GTC) to A (GCC) at codon 138 in exon 4, P (CCC) to S (TCC) at codon 151 in exon 5, and P (CCC) to L (CTC) at codon 151 in exon 5. Introduction of these mutations into the normal GALNS cDNA and transient expression in cultured fibroblasts resulted in a significant decrease in the enzyme activity. V138A and Q148X mutations result in changes of restriction site, which were analyzed by restriction-enzyme assay. P151S and P151L mutations that did not alter the restriction site were detected by direct sequencing or allele specific oligohybridization. Detection of the double gene deletion was initially done using Southern blots and was confirmed by PCR. Haplotypes were determined using seven polymorphisms to the GALNS locus in families with the double gene deletion. Haplotype analysis showed that the common double gene deletion occurred on a single haplotype, except for some variation in a VNTR-like polymorphism. This finding is consistent with a common founder for all individuals with this mutation.

5/3,AB/33 (Item 33 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

08872992 96226785 PMID: 8640101

Prevention of false results from preferential PCR amplification or VNTR alleles.

Pai CY; Chou SL; Tang TK; Wei YH; Yang CH

Department of Forensic Sciences, Central Police University, Taoyuan, Taiwan, ROC.

Journal of the Formosan Medical Association (TAIWAN) Jan 1996,

95 (1) p69-72, ISSN 0929-6646 Journal Code: BLQ

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

In forensic DNA typing, evidential samples generally involve limited amounts of DNA and so should be carefully utilized. Although polymerase chain reaction (PCR) of variable number of tandem repeats (VNTR) alleles is the prevailing method for forensic identification, the fidelity of amplification of heterozygous VNTR alleles with large disparities in length needs to be carefully examined. Reports in the literature and our own observations have demonstrated that PCR artifacts, bogus alleles and allelic drop-out of VNTRs, are related to the amount of genomic DNA, the number of amplification cycles and the

length of **alleles** amplified. Two small (< 1 kb) hypervariable VNTRs (Apo B and HVR-Ig) markers used for forensic identification were chosen to study these relationships. The results revealed that PCR amplification for the heterozygous VNTR **alleles** with wide disparity in length (> 400 bp) easily produced the allelic drop-out problem and therefore, led to the false results; and the allelic fragment of PCR products was preferentially lost after only 2 cycles of overamplification. We also further established the relationship between the optimal number of amplification cycles and the amount of genomic DNA in the reaction mixture. In our routine forensic screening this relationship has been successfully applied to determine the optimal number of amplification cycles and to avoid the allelic drop-out problem and achieve fidelity of PCR-VNTR amplification. It has also been used to investigate forensic casework.

5/3,AB/34 (Item 34 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

08868968 96207930 PMID: 8624036

PCR amplification of a polymorphic minisatellite VNTR locus in whiting (*Merlangius merlangus* L.).

McGregor D; Galvin P; Sadusky T; Cross T

Department of Zoology, University College Cork, Ireland.

Animal genetics (ENGLAND) Feb 1996, 27 (1) p49-51, ISSN 0268-9146 Journal Code: 4WE

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

An approach has been developed for the screening of allelic variation of minisatellite DNA loci that substantially reduces the time and hazards involved. Primers were designed for a minisatellite region isolated from a gadoid fish species (*Merlangius merlangus* L.), enabling amplification by polymerase chain reaction, so that differences in the number of minisatellite repeat units (allelic variability) were detectable by ethidium bromide fluorescence (over UV light) following separation by agarose gel electrophoresis. This amplifiable minisatellite variable number tandem repeat region, the first non-primate marker of its kind can be used successfully with DNA extracted by a rapid Chelex protocol. From a sample of 97 individuals, 24 **alleles** were resolved (750-2200 kb) and heterozygosity was estimated at 0.94.

5/3,AB/35 (Item 35 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

08865897 96172831 PMID: 8589723

Ovarian cancer risk in BRCA1 carriers is modified by the HRAS1 variable number of tandem repeat (VNTR) locus.

Phelan CM; Rebbeck TR; Weber BL; Devilee P; Rutledge MH; Lynch HT; Lenoir GM; Stratton MR; Easton DF; Ponder BA; Cannon-Albright L; Larsson C; Goldgar DE; Narod SA

Division of Medical Genetics, Montreal General Hospital, Quebec, Canada.

Nature genetics (UNITED STATES) Mar 1996, 12 (3) p309-11, ISSN 1061-4036 Journal Code: BRO

Contract/Grant No.: CA55914, CA, NCI

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

Women who carry a mutation in the BRCA1 gene (on chromosome 17q21), have an 80% risk of breast cancer and a 40% risk of ovarian cancer by the age of 70 (ref. 1). The variable penetrance of BRCA1 suggests that other genetic and non-genetic factors play a role in tumorigenesis in these individuals. The HRAS1 variable number of tandem repeats (VNTR) polymorphism,

located 1 kilobase (kb) downstream of the HRAS1 proto-oncogene (chromosome 11p15.5) is one possible genetic modifier of cancer penetrance. Individuals who have rare **alleles** of the VNTR have an increased risk of certain types of cancers, including breast cancer (2-4). To investigate whether the presence of rare HRAS1 **alleles** increases susceptibility to hereditary breast and ovarian cancer, we have typed a panel of 307 female BRCA1 carriers at this locus using a PCR-based technique. The risk for ovarian cancer was 2.11 times greater for BRCA1 carriers harbouring one or two rare HRAS1 **alleles**, compared to carriers with only common **alleles** ($P = 0.015$). The magnitude of the relative risk associated with a rare HRAS1 **allele** was not altered by adjusting for the other known risk factors for hereditary ovarian cancer (5). Susceptibility to breast cancer did not appear to be affected by the presence of rare HRAS1 **alleles**. This study is the first to show the effect of a modifying gene on the penetrance of an inherited cancer syndrome.

5/3,AB/36 (Item 36 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

08808630 96129347 PMID: 8586316

Practical application of three polymorphic microsatellites in intron 40 of the human von Willebrand factor gene.

Casana P; Martinez F; Aznar JA; Lorenzo JJ; Jorquera JJ

Unidad de Coagulopatias, Congeneticas de la Comunidad Valenciana, Espana.

Haemostasis (SWITZERLAND) Nov-Dec 1995, 25 (6) p264-71, ISSN

0301-0147 Journal Code: FYG

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

Intron 40 of the human von Willebrand factor gene contains a region with variable-number tandem repeats (VNTR), type (ATCT)_n, showing length polymorphism. In order to carry out family studies of von Willebrand's disease, we performed PCR procedures to analyze 3 previously described microsatellites from that region, both in normal individuals and in von Willebrand disease patients. Three pairs of primers were used to amplify independently nucleotides 1890-1991 (VNTR 1), 2215-2380 (VNTR 2) and 1640-1794 (VNTR 3) from intron 40. The observed heterozygosities (0.75, 0.73 and 0.86 for VNTRs 1, 2 and 3, respectively) were in good agreement with the expected heterozygosities derived from the **allele** frequencies (0.70, 0.73 and 0.79, respectively). Furthermore, the combination of the 3 VNTRs showed 96% of heterozygosity, which correspond with the 98% expected value under linkage equilibrium. Therefore, our conclusion is that the use of these 3 markers, especially VNTR 3, constitutes a rapid and reliable method for performing segregation studies in von Willebrand disease families.

5/3,AB/37 (Item 37 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

08799147 96056355 PMID: 7567434

St14 (DXS52) VNTR in the Chinese population and its application to genetic diagnosis of haemophilia A.

Wang X; Chu X; Ruan C

Jiangsu Institute of Hematology, Thrombosis and Hemostasis Research Unit, Suzhou Medical College, People's Republic of China.

Nouvelle revue francaise d'hematologie (GERMANY) 1995, 37 (3)

p183-6, Journal Code: O6S

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

The variable number of tandem repeats (VNTR) of St14 (DXS52) on the

human X-chromosome was analysed using the polymerase chain reaction (PCR) method. Screening of 78 X-chromosomes in 56 healthy Chinese individuals revealed the existence of at least seven different alleles in the Chinese population, the corresponding amplified fragments and frequencies being 700 bp (60.3%), 1220 bp (1.3%), 1300 bp (2.6%), 1390 bp (11.5%), 1570 bp (12.8%), 1630 bp (6.4%) and 1690 bp (5.1%). Total theoretical heterozygous rate was 60%. Compared to Caucasians, this Chinese population showed a markedly higher occurrence of low molecular weight fragments and a relatively low occurrence of high molecular weight fragments. Study of this polymorphism in 14 suspected haemophilia A carriers revealed half of them to be heterozygous. Thus, St14 VNTR analysis by PCR should prove to be a useful tool in the genetic diagnosis of haemophilia A in China.

5/3,AB/38 (Item 38 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

08778321 96329842 PMID: 8706151

[The variable number of tandem repeats in the renin gene of rats and possible significance in hypertension]

Zhang M; Fang F; Chen L

Institute of Basic Medical Sciences, CAMS, Beijing.

Zhongguo yi xue ke xue yuan xue bao (CHINA) Jun 1995, 17 (3)

p178-82, ISSN 1000-503X Journal Code: CZS

Languages: CHINESE

Document type: Journal Article

Record type: Completed

The variable number of tandem repeats (VNTR) in the first intron of renin gene for the spontaneously hypertensive rat (SHR), its controls Wistar-Kyoto (WKY), renal hypertensive rat, and Sprague-Dawley rat (SD) were compared by polymerase chain reaction (PCR) method. An analysis of VNTR from WKY, Wistar and SD showed that there are two different renin gene alleles and three genotypes 2.0kb/2.0kb, 2.0kb/1.8kb, 1.8kb/1.8kb. The genotype from renal hypertensive rats is same as those seen in the normal controls. However, compared with the WKY, Wistar and SD genes, a "deletion" of approximately 1.0kb was found in the first intron of the SHR renin gene. Our results strongly suggest that the cause and mechanism of elevated blood pressure is complex, and the molecular basis of the genetic-prone hypertension is existed.

5/3,AB/39 (Item 39 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

"nature" vs. "nurture"

08766461 95217437 PMID: 7702836

Highly discriminating heptaplex short tandem repeat PCR system for forensic identification.

Urquhart A; Oldroyd NJ; Kimpton CP; Gill P

Forensic Science Service, Birmingham, UK.

BioTechniques (UNITED STATES) Jan 1995, 18 (1) p116-8, 120-1,

ISSN 0736-6205 Journal Code: AN3

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

We describe a highly discriminating multiplex short tandem repeat PCR human identification system that gives a matching probability for Caucasians of European ancestry of 2.94×10^{-8} or 5.66×10^{-10} when used in combination with a previously described system. The system produces discrimination equal to or greater than four single locus probes (restriction fragment length polymorphism [RFLP] typing of variable nucleotide tandem repeat [VNTR] loci). The test is robust and reproducible and works with 1-10 ng of template DNA, using fluorescent detection of PCR products from either 4 or 6 short tandem repeat loci

and the X-Y homologous gene amelogenin, giving simultaneous sex diagnosis.

5/3,AB/40 (Item 40 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

08749141 96307986 PMID: 8703516

Amplified fragment length polymorphism of the VNTR locus COL2A1 in Chinese population]

Hou Y; Gou Q; Wu M

Department of Forensic Serology, West China University of Medical Sciences, Chengdu.

Yi chuan xue bao (CHINA) 1995, 22 (4) p245-51, ISSN 0379-4172
Journal Code: AO5

Languages: CHINESE

Document type: Journal Article

Record type: Completed

The amplifiable VNTR polymorphic system COL2A1 has been investigated in a Chinese Han population (n = 120) by the polymerase chain reaction (PCR) and PAGE horizontal electrophoresis followed by silver stain. In order to accurately identify COL2A1 alleles, a number of human allele ladders prepared by mixing DNAs extracted from different individuals of known COL2A1 genotypes were used. A total of 14 different alleles in 23 genotypes were observed in this Chinese Han population. Among them, four were new alleles disclosed in the present study. The results imply that COL2A1 locus may be served as a genetic marker in forensic haemogenetics as well as in anthropogenetics.

5/3,AB/41 (Item 41 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

08745545 96217002 PMID: 8650040

Evaluation of bone marrow grafts and hemopoietic chimerism using PCR hypervariable sequencing with variable number tandem repeat sequences]

Ocena przyjecia przeszczepu szpiku oraz chimeryzmu hemopoetycznego przy uzyciu amplifikacji metoda PCR hiperzmiennych sekwencji typu VNTR.

Zauchaj JM; Pawlowski R; Welz A; Prejzner W; Hauser R; Hellman A

Kliniki Hematologii Instytutu Chorob Wewnętrznych Akademii Medycznej w Gdansk.

Polski tygodnik lekarski (POLAND) Sep 1995, 50 (36-39) p73-4,
ISSN 0032-3756 Journal Code: PB5

Languages: POLISH

Document type: Journal Article

Record type: Completed

PCR amplification of highly polymorphic variable number of tandem repeat (VNTR) sequences could be particularly useful in documentation of engraftment and characterization of chimerism following allogeneic bone marrow transplantation (BMT). We have monitored a 31-year old male patient treated with allogeneic BMT for chronic myeloid leukaemia. The recipient's DNA samples were obtained before the transplant and on day 28, 100 and 150 after BMT. The donor's DNA (patient's sister) was also obtained as a reference. ACT B2 locus on chromosome 6 was chosen for the analysis. In addition a deletion polymorphism locus within the pseudoautosomal region of chromosome X and Y (amelogenin gene) was also analysed. On day 28 after BMT both donor and recipient specific alleles were detected in the recipient's sample. However, on day 100 and 150 the recipient specific alleles were no longer detectable. The aforementioned pattern was observed for both markers analysed. The disappearance of recipient specific alleles correlated with clinical symptoms of chronic graft-versus host disease.

5/3,AB/42 (Item 42 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

08745457 96213117 PMID: 8629128

DNA fingerprinting and forensic medicine.
Boonsaeng V
Department of Biochemistry, Faculty of Science Mahidol University,
Bangkok, Thailand.
Southeast Asian journal of tropical medicine and public health (THAILAND)
1995, 26 Suppl 1 p296-300, ISSN 0038-3619 Journal Code: UVN
Languages: ENGLISH
Document type: Journal Article; Review; Review, Tutorial
Record type: Completed

In forensic medicine, DNA fingerprinting for identification is becoming a necessary procedure. A method to radiolabel M13 DNA probe by primer extension using a specific oligonucleotide primer was developed. The method specifically labeled the two 15 bp repeats in M13 DNA which hybridize to target DNA giving rise to DNA fingerprinting patterns. The M13 probe labeled by this method has proven useful for individual identification, paternity testing and monitoring reconstitution in bone marrow transplantation. The genetic locus, D1S80 and D17S30 containing a variable number of tandem repeats (VNTR) have also been successfully amplified from human genomic DNA isolated from blood (50 ng from each sample) by the polymerase chain reaction (PCR) using oligonucleotide primers complementary to the flanking sequences as primers for amplification. DNA bands were detected by ethidium bromide staining after electrophoresis on agarose gels. Analysis of this VNTR locus was thus achieved without the need for Southern blot or radioactive material. The small size of the DNA fragments produced in the PCR amplification permitted good resolution of individual alleles. The precise specification of the number of tandem repeats present in each allelic fragment was reproducible from one analysis to another.

5/3,AB/43 (Item 43 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

08745032 96139975 PMID: 8586345

D1S80 VNTR locus genotypes in population of south Poland;
meta-analysis pointer to genetic disequilibrium of human populations.
Turowska B; Sanak M
Institute of Forensic Medicine, Faculty of Medicine, Jagiellonian
University, Krakow, Poland.
Forensic science international (IRELAND) Oct 30 1995, 75 (2-3)
p207-16, ISSN 0379-0738 Journal Code: F49
Languages: ENGLISH
Document type: Journal Article; Meta-Analysis
Record type: Completed

A highly variable number of tandem repeats (VNTRs) in a human locus D1S80 can prove to be useful for forensic science purposes. As with other genetic polymorphisms, a database of a local population allelic frequencies is needed to ensure that no departure from genetic equilibrium exists. DNA from the locus D1S80 was amplified by polymerase chain reaction (PCR) and analyzed by horizontal PAGE followed by silver staining. Samples from 133 unrelated inhabitants of Southern Poland were examined. The amplified fragment length polymorphism (AMP-FLP) analysis of the D1S80 locus demonstrated 21 alleles and heterozygosity of 0.85%. Out of the 231 possible genotypes, 47 were observed. The results were compared to the published D1S80 population studies and a meta-analysis of the genotype frequencies was performed. The G statistics revealed a deviation from genetic equilibrium in the Spanish population. Replicated goodness of fit tests showed highly significant heterogeneity of genotype distribution between tested populations. Therefore, interpretation of the casework on

the basis of D1S80 locus typing may be biased by interpopulation differences.

5/3,AB/44 (Item 44 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

08743763 96073030 PMID: 8536955

Polymorphism at **VNTR** locus 3 to the apolipoprotein B gene in a Tunisian population: difference from other ethnic groups.

Buresi C; Desmarais E; Vigneron S; Ben Rayana C; Chaabouni H; Roizes G
INSERM U 249, Montpellier, France.

Genetic epidemiology (UNITED STATES) 1995, 12 (4) p381-9,
ISSN 0741-0395 Journal Code: FMP

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

The Hypervariable region (HVR) detected at the 3' end of the apolipoprotein B (Apo B) locus has been the subject of numerous studies. As for many **VNTR** (variable number of tandem repeat), this locus is highly polymorphic and until now about 20 **alleles** have been described. The genotype distribution in all populations follows the Hardy-Weinberg predictions. A bimodal pattern of **allele** frequency distribution is apparent in all Caucasoid populations. We have analyzed the frequencies of different **alleles** in a Tunisian population (123 individuals) by the polymerase chain reaction technique and compared our results to those obtained in several ethnic groups. It appears that the distributions of the **allele** frequencies are very different: for Caucasoid populations, there are two peaks of frequencies for **alleles** with 36 and 48 repeats, but **alleles** of intermediate lengths are more frequent. Hixson et al. [(1993) Hum Genet 91:475-479] have shown a similar difference between black and white American populations. We found the same results in a black African group. Some of the repeat units of this HVR contain a Ssp I restriction site and digestion of the PCR products by this enzyme gives different patterns on gradient acrylamide gel [Desmarais et al., 1993, Nucleic Acids Res 21:2179-2184.] The DNA of African individuals (42) has been analyzed to discover the origin of this new **allele**. Preliminary results indicate that these particular **alleles** probably arose by introgression from the African population into the Tunisian one.

5/3,AB/45 (Item 45 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

08742506 96015359 PMID: 7479357

Detection of loss of heterozygosity in the p53 tumor suppressor gene using a PCR-based assay.

Ridanpaa M; Anttila S; Husgafvel-Pursiainen K

Department of Industrial Hygiene and Toxicology, Finnish Institute of Occupational Health, Helsinki, Finland.

Pathology, research and practice (GERMANY) Jun 1995, 191 (5)

p399-402, ISSN 0344-0338 Journal Code: PBZ

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

Inactivation of the p53 tumor suppressor gene has been reported to be a prognostic factor in several human cancer types. Normal function of the gene is affected by deletion in one **allele**; dysfunction of the other **allele** is often caused by a mutation. In tumors of heterozygous individuals, deletion of one **allele** can be detected as loss of heterozygosity (LOH). A recently found variable number of tandem repeats (**VNTR**) segment in intron 1 of the p53 gene seems to be highly polymorphic and, therefore, a very useful marker in detecting LOH in

various types of tumor samples. We in vitro amplified the VNTR segment from genomic DNA samples of 101 lung cancer patients and run conventional agarose gel electrophoreses in order to detect the alleles of various length, differing by the number of repeats. The usefulness of the method was studied using DNA from white blood cell samples and from fresh and formalin-fixed, paraffin-embedded tumor samples. Of the patients, 56% were found to have two different alleles, i.e. were informative in this assay. In 18% of the lung tumors from the informative cases, LOH in the p53 suppressor gene was detected.

5/3,AB/46 (Item 46 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

08742406 96016915 PMID: 7476773

[A polymorphism study of the DNA extracted from dental tissues]
Studio di polimorfismi del DNA estratto da tessuti dentari.
Dell'Osso G; Avitabile M; Sciacca G; Lanteri E; Stivala F
Istituto di Medicina Legale e delle Assicurazioni, Università degli Studi, Catania.

Minerva stomatologica (ITALY) May 1995, 44 (5) p205-9, ISSN 0026-4970 Journal Code: NB2

Languages: ITALIAN

Document type: Journal Article

Record type: Completed

Often teeth are the only items which can be used for personal identification in forensic medicine. In the present work we describe a method to extract and amplify DNA from dental elements ranging from 2 weeks to 5 year from the avulsion. PCR (polymerase chain reaction) was used to amplify VNTR sequences; the alleles products were electrophoresed, visualized by traditional methods and compared to the amplified products obtained from the matching blood sample. Our results give a new and powerful investigative tool for personal identification in the field of forensic odontostomatology, since such a procedure can be successfully applied both to recent and to ancient teeth.

5/3,AB/47 (Item 47 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

08741663 96090095 PMID: 7580849

Rapid typing of 4 VNTR loci, 3'ApoB, MCT118, St14 and YNZ22 by the polymerase chain reaction of a Greek sample.

Lambropoulos AF; Frangoulides E; Kotsis A; Dozi-Vassiliades I
Aristotle University of Thessaloniki, Medical Faculty, Dept. of General Biology, Greece.

Cellular and molecular biology (FRANCE) Jul 1995, 41 (5) p699-702, ISSN 0145-5680 Journal Code: BNA

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

Allelic data from a Greek sample of unrelated individuals for the D17S30, 3'ApoB, D1S80 and DXS52 loci were obtained by the PCR and subsequent analysis with agarose gel electrophoresis. The distribution of observed genotypes is in agreement with expected values according to the Hardy-Weinberg equilibrium. An heterozygosity of at least 76% was demonstrated with 11-13 alleles for each locus. The use of the combination of the four loci could be a powerful tool for paternity testing and individual discrimination, in combination with the fact that PCR needs a minimal amount of DNA and is a very quick and sensitive method.

5/3,AB/48 (Item 48 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

08740653 96057835 PMID: 7551967

Association of polymorphic **VNTR** region in the first intron of the human TH gene with disturbances of the catecholamine pathway in schizophrenia.

Wei J; Ramchand CN; Hemmings GP

Institute of Biological Psychiatry, Schizophrenia Association of Great Britain, Bryn Hyfryd, The Crescent, Bangor, Gwynedd, UK.

Psychiatric genetics (ENGLAND) Summer 1995, 5 (2) p83-8,
ISSN 0955-8829 Journal Code: B3X

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

In the present study, five allelic fragments were typed by a polymerase chain reaction (PCR) process with a pair of primers specific for the tetranucleotide (TCAT) repeat sequence in the first intron of the human tyrosine hydroxylase (TH) gene and their sizes (bp) were 114 (A), 118 (B), 122 (C), 126 (D) and 130 (E), respectively. The AE genotypic frequency was found to be significantly higher in unrelated patients with schizophrenia than in unrelated control subjects ($\chi^2 = 4.18$, $p < 0.05$). ANOVA revealed a significant difference between the three groups (neuroleptic-free patients possessing or not possessing the AE genotype, and unrelated control subjects) in the concentration of serum noradrenaline ($F = 4.96$, $df = 2.79$, $p < 0.01$), but no significant differences were found between the three groups in the concentrations of serum homovanillic acid, phenylalanine and tyrosine. These results suggest that the polymorphic intron 1 of the human TH gene may be associated with disturbances of the catecholamine pathway in schizophrenia.

5/3,AB/49 (Item 49 from file: 155)

DIALOG(R) File 155:MEDLINE(R)

08739587 95375255 PMID: 7647320

The association of polymorphisms at a **VNTR** locus 3' to the apolipoprotein B gene with coronary heart disease in Chinese population.

Ye P; Chen B; Wang S

Chinese PLA General Hospital, Beijing.

Chinese medical sciences journal (CHINA) Jun 1995, 10 (2)
p63-9, ISSN 1001-9294 Journal Code: A8E

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

The polymorphisms of variable number of tandem repeats (**VNTR**) 3' to the apolipoprotein B (apo B) gene were investigated using polymerase chain reaction (PCR) in a sample of 103 patients with documented coronary heart disease (CHD) and 100 healthy individuals selected from Chinese Han nationality. Twelve segregating **alleles** (3' beta 29-51) were observed in the pooled total of 203 subjects. The most common **allele** was 3' beta 37, followed by 3' beta 39 with frequencies of 0.362 and 0.296, respectively. This model of **allele** distribution was coincident with the results from different ethnic groups, but the relative frequencies of **alleles** were different. In comparison with the **allele** frequencies between the patients and controls, **alleles** bigger than 3' beta 39 (3' **VNTR** -B) were significantly more common among the patients than among the controls ($P < 0.001$). Moreover, in the CHD group patients with plasma levels of TC $> \text{or} = 3.88$ mmol/L, LDL-C $> \text{or} = 2.59$ mmol/L and HDL-C < 1.16 mmol/L had significantly higher frequencies of 3' **VNTR**-B **allele** ($P < 0.01$). Therefore, it is suggested that 3' **VNTR**-B **allele** might be involved in the development of coronary atherosclerosis, presumably through their influences on lipid metabolism.

5/3,AB/50 (Item 50 from file: 155)

DIALOG(R) File 155:MEDLINE(R)

08738138 95329953 PMID: 7606177

Frequency distribution of hypervariable VNTRs in Apo B, HVR-Ig and COL2A1 loci in Taiwan: forensic application.

Pai CY; Yang CH; Chou SL; Wei YH

Department of Forensic Science, Central Police University, Taoyuan, Taiwan ROC.

Journal of the Formosan Medical Association (TAIWAN) Apr 1995,

94 (4) p164-71, ISSN 0929-6646 Journal Code: BLQ

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

By use of a simple, rapid and reliable polymerase chain reaction (PCR)-based method, we analyzed three hypervariable tandem repeats in the 3'-Apo B, 5'-HVR-Ig and 3'-COL2A1 loci. As accurate data of allele frequency of genetic markers is a prerequisite for forensic application, the allele frequency distribution of the three variable number of tandem repeats (VNTR) among the Chinese population in Taiwan were studied. In a total of 123 unrelated Chinese subjects, the Apo B VNTR demonstrated a heterozygosity of 68.2% with 9 alleles, 0.85 of the power of discrimination (PD) value and 0.74 of the allelic diversity (h) value. In a sample of 103 unrelated Chinese subjects, the COL2A1 VNTR showed 49.0% heterozygosity with six alleles, 0.79 of the PD value and 0.74 of the h value. In 106 unrelated subjects, the HVR-Ig VNTR showed 47.4% heterozygosity with seven alleles, 0.79 of the PD value and 0.59 of the h value. The data obtained in this study are not only useful for forensic identification, but will also be helpful for paternity testing, genetic linkage studies and the identification of the three VNTR loci associated with human genetic diseases. Some verifying examinations for the validity and reliability of the three VNTR were performed. The high sensitivity and inexpensive nature of this approach make it superior to the traditional method of DNA fingerprinting for forensic typing. With the use of this PCR-VNTR system, many forensic cases have been successfully identified. The value of this system is illustrated in the investigation of a rape and murder case.

5/3,AB/51 (Item 51 from file: 155)

DIALOG(R) File 155:MEDLINE(R)

08738020 95325814 PMID: 7602291

French Caucasian population data for HUMTH01 and HUMFES/FPS short tandem repeat (STR) systems.

Pfitzinger H; Ludes B; Kintz P; Tracqui A; Mangin P

Institut de Medecine Legale, Faculte de Medecine, Strasbourg, France.

Journal of forensic sciences (UNITED STATES) Mar 1995, 40 (2)

p270-4, ISSN 0022-1198 Journal Code: I5Z

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

The recent technology of amplification of DNA sequences by the polymerase chain reaction (PCR) has already proved to be a very useful tool for the analysis of variable number of tandem repeat (VNTR) loci. Short tandem repeat (STR) loci appear as other promising PCR-based identification systems. In fact, DNA typing based on PCR amplification of STRs is very sensitive and allows to overcome major problems encountered when using the RFLP method, such as typing of very small amounts of DNA, highly degraded DNA or mixtures of DNA from more than one individual. Two STR systems, HUMTH01 (a tetranucleotide repeat (AATG) sequence located on chromosome 11) and HUMFES/FPS (a tetranucleotide repeat (ATTT) sequence located on chromosome 15) were investigated in order to determine allele and genotype frequencies for a French caucasian

population sample. HUMTH01 and HUMFES/FPS alleles were amplified by the use of PCR and amplified STR sequences were analyzed on 6% Hydrolink Long Ranger gels and visualized by silver staining. The study was conducted on a sample of unrelated individuals (N approximately 190) randomly selected from the French caucasian population. The genotype distributions met Hardy-Weinberg expectations for both HUMTH01 and HUMFES/FPS STR systems. Furthermore, an additional allele, never reported before was observed at the HUMFES/FPS locus: it migrates as an allele containing 7 repeat units and corresponds to the smallest allele identified for this locus.

5/3,AB/52 (Item 52 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

08737901 95322361 PMID: 7599103

Suitability of the YNZ22 (D17S5) VNTR polymorphism for legal medicine investigations in the population of Catalonia (Spain).

Gene M; Huguet E; Sanchez-Garcia C; Moreno P; Corbella J; Mezquita J
Department of Legal Medicine, Faculty of Medicine, University of Barcelona, Spain.

International journal of legal medicine (GERMANY) 1995, 107 (4)

p222-4, ISSN 0937-9827 Journal Code: AX1

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

Allele and phenotype frequencies for the YNZ22 locus were determined in a population sample from Catalonia (Spain) using the polymerase chain reaction (PCR). In 311 unrelated individuals, 14 alleles and 56 phenotypes were observed. No deviation from Hardy-Weinberg equilibrium was found. The observed heterozygosity was 81.35%. The YNZ22 polymorphism is useful for paternity testing with a CE value of 70% and an Essen-Moller value of 9.35 (log.).

5/3,AB/53 (Item 53 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

08735127 95221217 PMID: 7706114

Population structure, stepwise mutations, heterozygote deficiency and their implications in DNA forensics.

Jin L; Chakraborty R

Center for Demographic and Population Genetics, Graduate School of Biomedical Sciences, University of Texas, Houston 77225.

Heredity (ENGLAND) Mar 1995, 74 (Pt 3) p274-85, ISSN

0018-067X Journal Code: G6N

Contract/Grant No.: GM 41399, GM, NIGMS; GM 45861, GM, NIGMS

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

In a substructured population the overall heterozygote deficiency can be predicted from the number of subpopulations (s), their time of divergence (t), and the nature of the mutations. At present the true mutational mechanisms at the hypervariable DNA loci are not known. However, the two existing mutation models (the infinite allele model (IAM) and the stepwise mutation model (SMM)) provide some guides to predictions from which the possible effect of population substructuring may be evaluated, assuming that the subpopulations do not exchange any genes among them during evolution. The theory predicts that the loci with larger mutation rate, and consequently showing greater heterozygosity within subpopulations, should exhibit a smaller proportional heterozygote deficiency (GST) and, hence, the effects of population substructuring should be minimal at the hypervariable DNA loci (an order of magnitude smaller than that at the blood group and protein loci). Applications of

this theory to data on six Variable Number of Tandem Repeat (VNTR) loci and five short tandem repeat (STR) loci in the major cosmopolitan populations of the USA show that while the VNTR loci often exhibit a large significant heterozygote deficiency, the STR loci do not show a similar tendency. This discordant finding may be ascribed to the limitations, coalescence and nondetectability of alleles associated with the restriction fragment length polymorphism (RFLP) analysis through which the VNTR loci are scored. Such limitations do not apply to the polymerase chain reaction (PCR) method, through which the STR loci are scored. The implications of these results are discussed in the context of the forensic use of DNA typing data.

5/3,AB/54 (Item 54 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

08735077 95218198 PMID: 7703498

Quantitative determination of bone marrow transplant engraftment using fluorescent polymerase chain reaction primers for human identity markers.

Scharf SJ; Smith AG; Hansen JA; McFarland C; Erlich HA

Department of Molecular Genetics, Roche Molecular Systems, Inc, Alameda, CA 94501, USA.

Blood (UNITED STATES) Apr 1 1995, 85 (7) p1954-63, ISSN 0006-4971 Journal Code: A8G

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

We have developed a quantitative, nonisotopic method using variable number tandem repeat (VNTR) and short tandem repeat (STR) markers for monitoring donor cell engraftment in marrow transplant recipients. Posttransplant DNA from the recipient is amplified with fluorescent polymerase chain reaction (PCR) primers for polymorphic markers that distinguish donor alleles from recipient alleles. The fluorescent PCR products are then separated on agarose or acrylamide gels on the Applied Biosystems 373A Sequencer (Foster City, CA). Using GeneScan 672 software (Applied Biosystems) to analyze the separated alleles, we can correlate allele peak areas to the percentage of donor or recipient DNA. We quantitate engraftment in a mixed chimeric sample by mixing pretransplant recipient and donor DNAs in a range of percentages and amplifying the mixtures to produce a standard curve. By amplifying and analyzing the posttransplant sample DNA(s), we can determine the extent of engraftment by interpolating the percent peak area of the informative allele(s) from this standard curve. This approach provides a precision of measurement ranging, depending on the marker, from 3.5% to 8.0% (percent coefficient of variation) and an accuracy of engraftment determination ranging from 97% to 99%, with a sensitivity of detection of 1% donor or recipient DNA. We retrospectively analyzed a panel of 32 patients and found seven to be informative for some degree of mixed chimerism, indicative of either residual normal host cells or leukemic relapse. An analysis of different cell lineages obtained posttransplant showed different degrees of engraftment in myeloid and T-cell populations. In summary, this method can provide an accurate, quantitative assessment of mixed chimerism in patients posttransplant. Such information may be useful in the future in guiding early implementation of additional treatment designed to circumvent graft failure or suppress relapse.

5/3,AB/55 (Item 55 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

08734368 95172467 PMID: 7868010

Allele frequency distribution of four PCR-amplified loci in the Spanish population.

Cabrero C; Diez A; Valverde E; Carracedo A; Alemany J

PharmaGen, Madrid, Spain.
Forensic science international (IRELAND) Jan 30 1995, 71 (2)
p153-64, ISSN 0379-0738 Journal Code: F49
Languages: ENGLISH
Document type: Journal Article
Record type: Completed

The **allele** frequency distributions of four **VNTR** loci amplified by **PCR** have been studied in a population of 205 individuals from Spain. The loci analysed are D1S80 and three STRs: HUMTH01, HUMFES/FPS and HUMACTBF2 (SE33). The former was visualized in Metaphor agarose gels, and the STRs in sequencing polyacrylamide gels under denaturing conditions which could separate **alleles** with differences of a single base. This is of particular importance in the HUMTH01 locus, a tetrameric STR in which two **alleles** (9.3 and 10) were detected differing in a single base. Furthermore, HUMACTBP2 has at least 30 **alleles**, some of which may vary by as little as one base. At this locus a variation in the **allele** mobility was observed, depending on the electrophoretic conditions. For this reason, there should be careful consideration before this marker is accepted and validated as a common interlaboratory system. This paper does not include any comparison of the frequencies obtained for this locus with other recent studies. For the rest of the loci, the frequencies found have been compared with other published population studies; they show a degree of difference, particularly in the D1S80 locus. Finally, the systems were tested for Hardy-Weinberg equilibrium, and some statistical parameters of forensic interest were calculated.

5/3,AB/56 (Item 56 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

08694869 96144796 PMID: 8550242

Detection of p53 gene alteration in renal-cell cancer by micropreparation techniques of tumor specimens.

Kuczyk MA; Serth J; Bokemeyer C; Jonassen J; Arndt H; Paeslack U; Werner M; Tan HK; Jonas U

Department of Urology, Hanover University Medical School, Germany.

International journal of cancer. Journal international du cancer (UNITED STATES) Dec 20 1995, 64 (6) p399-406, ISSN 0020-7136

Journal Code: GQU

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

Alterations in the p53 tumor-suppressor gene have been identified in a variety of human malignancies, including renal-cell cancer. A technique for the isolation of tumor areas from tissue specimens to analyze formalin-fixed and paraffin-embedded tumors and try to avoid a disturbance of the results due to genetic background signal by the presence of tumor-infiltrating lymphocytes, was established. The presence of lymphocytes within the tumor areas investigated was determined by immunohistochemical staining for CD3, a lymphatic surface antigen. Following the isolation of about 100-200 tumor cells, **PCR**-directed molecular genetic analysis was performed. A highly informative allelotyping approach for the detection of loss of heterozygosity (LOH), determining BstU1- and **VNTR**-polymorphisms, a 100-bp marker directly localized in intron 1 of the p53 gene, as well as screening for mutations by single-strand conformation polymorphism analysis (SSCP) in exons 5-8, were used. Out of 44 renal-cancer specimens, 33 (75%) were informative for **PCR**-directed RFLP-analysis. Allelic loss at the p53 gene locus was observed in 10 of 33 cases (33%). No correlation between p53 gene alteration and T-stage, histological grade or histological differentiation could be observed. Alterations in the p53 gene, as detected by a molecular genetic as well as an immunohistochemical approach, were correlated to overall survival. During univariate analysis histological grade, lymphnode status and the presence of distant metastases could be identified as

prognostic parameters for overall survival. During multivariate analysis none of the factors investigated remained an independent prognosticator for survival. Summarizing these results, it seems unlikely that p53 gene alterations will serve as an important new factor for the clinical prognosis of patients with renal-cell cancer.

5/3,AB/57 (Item 57 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

08693005 96122101 PMID: 8547165

Observation of null **alleles** apparently due to deletions.

Moller A; Wiegand P; Brinkmann B

Institute of Legal Medicine, Westfalische Wilhelms-Universitat, Munster, Germany.

International journal of legal medicine (GERMANY) 1995, 108 (2)

p90-2, ISSN 0937-9827 Journal Code: AX1

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

After examining 2 paternity cases in 17 classical, 4 RFLP and 5 **PCR**-**VNTR** systems, isolated pseudo-exclusions were observed in the polymorphism D2S44 (YNH24). In both cases the "exclusions" were due to apparent opposite homozygosity. The application of different restriction enzymes, **PCR** amplification and varying electrophoretic conditions each led to an equivalent result of a 1-band-pattern with a mismatch between both father/child pairs. From these results the authors conclude that a complete or almost complete loss of the **alleles** is the most probable explanation.

5/3,AB/58 (Item 58 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

08692717 96037023 PMID: 8546754

Association of polymorphisms of the apolipoprotein B gene with coronary heart disease in Han Chinese.

Ye P; Chen B; Wang S

Division of Geriatric Cardiology, Chinese PLA General Hospital, Beijing, China.

Atherosclerosis (IRELAND) Sep 1995, 117 (1) p43-50, ISSN

0021-9150 Journal Code: 95X

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

Four polymorphic sites of the apolipoprotein B (apo B) gene were investigated by using polymerase chain reaction (**PCR**) in 103 patients with coronary heart disease (CHD) and 100 age-matched healthy individuals selected from a population of Han Chinese in the Beijing area. The rare X+ **allele** of the XbaI restriction site was more frequently seen in CHD patients than in controls (0.088 vs. 0.025, $P < 0.01$). The relative frequency of rare E- **allele** of the EcoRI restriction site was significantly higher in CHD patients compared with controls (0.11 vs. 0.04, $P < 0.01$). Similarly, 3'**VNTR**-L **allele** (number of repeat units > 39) at the **VNTR** region was also present at an apparently high frequency in CHD patients in comparison to that in controls (0.602 vs. 0.290, $P < 0.001$). However, the difference in relative frequency of rare Del **allele** of the Ins/Del polymorphism at the signal peptide was not significant between the two groups (0.282 vs. 0.235, $P > 0.05$). In comparison with Caucasians, the relative frequencies of rare **alleles** (Del, X+ and E-) were found to be statistically lower in Han Chinese. Furthermore, the Del and X+ **alleles**, in linkage disequilibrium, were associated with significantly lower plasma level of HDL-C in CHD patients. Therefore it is suggested that genetic variation with the apo B gene may

exert some impact on lipid metabolism and contribute to the susceptibility to development of CHD in Han Chinese.

5/3,AB/59 (Item 59 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

08681331 96089456 PMID: 8529610

Rapid typing of variable number of tandem repeat locus in the human apolipoprotein B gene for DNA diagnosis of heart disease by polymerase chain reaction and capillary electrophoresis.

Baba Y; Tomisaki R; Sumita C; Morimoto I; Sugita S; Tsuhako M; Miki T; Ogihara T

Department of Chemistry, Kobe Pharmaceutical University, Japan.

Electrophoresis (GERMANY) Aug 1995, 16 (8) p1437-40, ISSN

0173-0835 Journal Code: ELE

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

The apolipoprotein B (apoB) variable number of tandem repeat (VNTR) alleles containing larger repeat units is a risk factor for coronary heart disease. Capillary electrophoresis (CE) in entangled polymer solution was applied to the analysis of polymerase chain reaction (PCR) amplified apoB VNTR locus for DNA diagnosis of heart disease. The CE separation gives an excellent resolution of two alleles differing by one or two 16 bp repeat units in the DNA size range up to 600 bp with high speed. The apoB alleles differing in length by 2 or 4 repeat units are readily distinguishable by CE in the DNA size range from 600 to 1000 bp. The plate number achieved was 1 million plates per meter. CE combining with PCR provides an excellent technique for accurate determination of the number of repeat units of apoB VNTR alleles and differentiation of heterozygous from homozygous individuals. Using the CE technique, the apoB VNTR loci from some individuals in genotyping were examined towards precise DNA diagnosis for coronary heart disease.

5/3,AB/60 (Item 60 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

08673759 95375431 PMID: 7647463

Evolution of a repeat sequence in the parathyroid hormone-related peptide gene in primates.

Pausova Z; Morgan K; Fujiwara TM; Hendy GN

Department of Medicine, McGill University, Montreal, Quebec, Canada.

Mammalian genome (UNITED STATES) Jun 1995, 6 (6) p408-14,

ISSN 0938-8990 Journal Code: BES

Erratum in Mamm Genome 1995 Sep;6(9) 691

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

A polymorphism of the variable number of tandem repeat (VNTR) type is located 97 bp downstream of exon VI of the parathyroid hormone-related peptide (PTHrP) gene in humans. The repeat unit has the general sequence G(TA)_nC, where n equals 4-11. In order to characterize the evolutionary history of this VNTR, we initially tested for its presence in 13 different species representing four main groups of living primates. The sequence is present in the human, great apes, and Old World monkeys, but not in New World monkeys; and this region failed to PCR amplify in the Loris group. Thus, the evolution of the sequence as part of the PTHrP gene started at least 25-35 millions years ago, after divergence of the Old World and New World monkeys, but before divergence of Old World monkeys and great apes and humans. The structural changes occurring during evolution are characterized by a relatively high degree of sequence divergence. In general, the tandem repeat region tends to be longer and more complex in

higher primates with the repeat unit motifs all being based on a TA-dinucleotide repeat sequence. Intra-species variability of the locus was demonstrated only in humans and gorilla. The divergence of the TA-dinucleotide repeat sequence and the variable mutation rates observed in different primate species are in contrast to the relative conservation of the flanking sequences during primate evolution. This suggests that the nature of the TA-dinucleotide repeat sequence, rather than its flanking sequences, is responsible for generating variability. (ABSTRACT TRUNCATED AT 250 WORDS)

5/3,AB/61 (Item 61 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

08650151 96080965 PMID: 7595333

Arab population data on the PCR-based loci: HLA-DQA1, LDLR, GYPA, HBGG, D7S8, Gc, and D1S80.

Hayes JM; Budowle B; Freund M
Department of Forensic Sciences, George Washington University,
Washington, DC, USA.

Journal of forensic sciences (UNITED STATES) Sep 1995, 40 (5)
p888-92, ISSN 0022-1198 Journal Code: I5Z

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

Allele and genotype frequencies for seven polymerase chain reaction (PCR)-based DNA genetic markers were determined in an Arab sample population. The loci analyzed were HLA-DQA1, LDLR, GYPA, HBGG, D7S8, Gc and D1S80. Results were obtained from the first six loci using the AmpliType HLA-DQ alpha DNA and AmpliType PM PCR Amplification and Typing Kits. The **VNTR** locus D1S80 PCR product was analyzed by polyacrylamide electrophoresis and silver staining. All loci meet Hardy-Weinberg expectations. The frequency data can be used in forensic analyses and paternity tests to estimate the frequency of a DNA profile in the Arab population.

5/3,AB/62 (Item 62 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

08645965 96026180 PMID: 7589827

Insulin gene 5' flanking polymorphism. Length of class 1 **alleles** in number of repeat units.

McGinnis RE; Spielman RS
Department of Genetics, University of Pennsylvania School of Medicine,
Philadelphia 19104-6145, USA.

Diabetes (UNITED STATES) Nov 1995, 44 (11) p1296-302, ISSN
0012-1797 Journal Code: E8X

Contract/Grant No.: DK-46618, DK, NIDDK

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

The 5' flanking polymorphism (5'FP) is a minisatellite, variable number of tandem repeat (**VNTR**) locus adjacent to the 5' end of the insulin gene (INS). **Alleles** of the 5'FP are highly variable in length but fall into three discrete size classes. The shortest, or class 1, **alleles** are associated with insulin-dependent diabetes mellitus (IDDM). Here we present a polymerase chain reaction (PCR)-based technique for subtyping 5'FP class 1 **alleles** by determining their exact lengths in number of repeat units (RUs). The technique resolves small length differences not detectable by Southern blot and produces a frequency distribution of class 1 **allele** lengths, which serve as subtypes of the crude class 1 category. We have applied the technique to 132 Caucasian families with IDDM offspring and have found that the lengths of 5'FP class

1 **alleles** form a quasi-continuous distribution with three distinct modes. We also found precise correlation between class 1 **allele** length and the **allele** present on the same chromosome at HUMTH01, a second **VNTR** locus in the INS region. Specifically, each of the four common **alleles** of HUMTH01 exhibited near-total association with a narrow size range belonging to one of the three components of the class 1 distribution. We discuss these results in relation to the population history of the 5'FP and INS region haplotypes and in relation to IDDM susceptibility in the INS region.

5/3,AB/63 (Item 63 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

08622087 96018181 PMID: 7557351
Population genetic study of the human dopamine transporter gene (DAT1).
Doucette-Stamm LA; Blakely DJ; Tian J; Mockus S; Mao JI
A Division of Genome Therapeutics Corporation, Waltham, Massachusetts
02154, USA.

Genetic epidemiology (UNITED STATES) 1995, 12 (3) p303-8,
ISSN 0741-0395 Journal Code: FMP

Contract/Grant No.: R44GM44454-02, GM, NIGMS

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

The human dopamine transporter gene, DAT1, acts to transport released dopamine into presynaptic terminals of the brain. The possibility that the DAT1 gene plays a role in genetic diseases of the brain has led to studies of DAT1 in several psychiatric and neurological disorders. Previous sequence analysis of DAT1 revealed a 40-bp repeat in the 3' end of the gene. In order to identify all potential **alleles** for this **VNTR** marker a population database was established. One thousand seventy-four unrelated individuals were screened by PCR for the region containing the 40 bp repeat. **Allele** frequency differences were found between black Americans and Caucasians or Hispanics but no differences were observed between Caucasians and Hispanics. A previously unreported **allele** was detected in all three populations. Thus, we have shown that screening a large population identifies new **alleles** and generates more accurate **allele** frequencies.

5/3,AB/64 (Item 64 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

08538707 95308348 PMID: 7788550
[Expression and loss of heterozygosity of DCC gene in human lung cancer]
Zhang J; Ding F; Wang X
National Laboratory of Molecular Oncology, Chinese Academy of Medical
Science, Beijing.

Zhonghua yi xue za zhi (CHINA) Apr 1995, 75 (4) p211-3, 254-5,
ISSN 0376-2491 Journal Code: CDG

Languages: CHINESE

Document type: Journal Article

Record type: Completed

The level of DCC mRNA expression was evaluated in tissue specimens from lung cancer patients by semiquantitative reverse transcription-polymerase chain reaction (RT-PCR) combined with Southern blot analysis. Obvious reduction of DCC gene expression was observed in 4 of 7 specimens (55%). In two specimens DCC transcript could only be detected after Southern blot hybridization of RT-PCR product. The average level of DCC expression in cancer tissue was about 45% of normal tissue as estimated by laser densitometer. We also studied DNA samples for loss of heterozygosity (LOH) at DCC locus at two polymorphic sites. Among the 15 specimens including 7 samples for RT-PCR, 9 (60%) were informative at either of two

polymorphic sites. LOH was observed in 5 (55%). Two at the MspI-RFLP (restriction fragment length polymorphism) site and 3 at the site of VNTR (variable number of tandem repeat). These results suggest that allele loss and decreased expression of DCC gene are frequent events and the possible involvement of DCC gene in the pathogenesis of human lung cancer.

5/3,AB/65 (Item 65 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

08466277 95150028 PMID: 7847372

An RNA-splicing mutation (G+5IVS20) in the type II collagen gene (COL2A1) in a family with spondyloepiphyseal dysplasia congenita.

Tiller GE; Weis MA; Polumbo PA; Gruber HE; Rimoin DL; Cohn DH; Eyre DR
Department of Pediatrics, Vanderbilt University School of Medicine, Nashville.

American journal of human genetics (UNITED STATES) Feb 1995, 56
(2) p388-95, ISSN 0002-9297 Journal Code: 3IM
Contract/Grant No.: AR01925, AR, NIAMS; AR37318, AR, NIAMS; HD22657, HD, NICHD

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

Defects in type II collagen have been demonstrated in a phenotypic continuum of chondrodysplasias that includes achondrogenesis II, hypochondrogenesis, spondyloepiphyseal dysplasia congenita (SEDC), Kniest dysplasia, and Stickler syndrome. We have determined that cartilage from a terminated fetus with an inherited form of SEDC contained both normal alpha 1(II) collagen chains and chains that lacked amino acids 256-273 of the triple-helical domain. PCR amplification of this region of COL2A1, from genomic DNA, yielded products of normal size, while amplification of cDNA yielded a normal sized species and a shorter fragment missing exon 20. Sequence analysis of genomic DNA from the fetus revealed a G-->T transversion at position +5 of intron 20; the affected father was also heterozygous for the mutation. Allele-specific PCR and heteroduplex analysis of a VNTR in COL2A1 independently confirmed the unaffected status of a fetus in a subsequent pregnancy. Thermodynamic calculations suggest that the mutation prevents normal splicing of exon 20 by interfering with binding of U1 small-nuclear RNA to pre-mRNA, thus leading to skipping of exon 20 in transcripts from the mutant allele. Electron micrographs of diseased cartilage showed intracellular inclusion bodies, which were stained by an antibody to alpha 1(II) procollagen. Our findings support the hypothesis that alpha-chain length alterations that preserve the Gly-X-Y repeat motif of the triple helix result in partial intracellular retention of alpha 1(II) procollagen and produce mild to moderate chondrodysplasia phenotypes.

5/3,AB/66 (Item 66 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

08450182 95169239 PMID: 7865100

Genetic association between dopamine transporter protein alleles and cocaine-induced paranoia.

Gelernter J; Kranzler HR; Satel SL; Rao PA
Department of Psychiatry, Yale University School of Medicine, West Haven, Connecticut.

Neuropsychopharmacology (UNITED STATES) Nov 1994, 11 (3)
p195-200, ISSN 0893-133X Journal Code: ADQ
Contract/Grant No.: DA04060, DA, NIDA; DA05592, DA, NIDA; K20-MH00931, MH, NIMH; +

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

Paranoia in the context of cocaine abuse is common and potentially dangerous. Several lines of evidence suggest that this phenomenon may be related to function of the dopamine transporter protein (DAT). DAT is the site of presynaptic reuptake of dopamine, an event that terminates its synaptic activity. The gene coding for dopamine transporter protein (DAT1) contains a variable number of tandem repeats (VNTR) polymorphism in the 3' untranslated region that can be typed by the polymerase chain reaction (PCR) (Vandenberg et al. 1992). Although this is not a coding region polymorphism, it is close to the coding region and could plausibly be in linkage disequilibrium with a mutation in the gene. Cocaine blocks the dopamine transporter and increases synaptic availability of dopamine. We examined DAT alleles in 58 white and 45 black cocaine users in order to test only two hypotheses: (1) Is there an allelic association between DAT and cocaine dependence? and (2) Is there an allelic association between DAT and cocaine-induced paranoia? We did not demonstrate an allelic association with cocaine dependence. However, within the white sample, DAT genotype was associated with cocaine-induced paranoia (allele frequency for allele 9 = .16 for those without paranoid experiences versus .35 for those with, $\chi^2 = 3.9$ [2 x 2 table], $p < .05$). There was no significant difference for the same measure in the black sample. Certain DAT genotypes may therefore predispose to paranoia in the context of cocaine use in white populations. We caution that these results require independent replication.

5/3,AB/67 (Item 67 from file: 155)

DIALOG(R) File 155:MEDLINE(R)

08419386 95004026 PMID: 7920240

Characterization of human AFLP systems apolipoprotein B, phenylalanine hydroxylase, and D1S80.

Latorra D; Stern CM; Schanfield MS

Roche Diagnostic Systems, Inc., Branchburg, New Jersey 08876.

PCR methods and applications (UNITED STATES) Jun 1994, 3 (6)

p351-8, ISSN 1054-9803 Journal Code: BNV

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

Methodology is presented for amplified fragment length polymorphism (AFLP) typing using a nonisotopic, PCR protocol. Human variable number tandem repeat (VNTR) loci used for identification in forensic and paternity testing were optimized for reaction and thermal-cycling parameters. Loci analyzed were the apolipoprotein B (APOB) 3' hypervariable region (HVR), phenylalanine hydroxylase 3' HVR (PAH), and D1S80. Coamplification of a monomorphic beta-globin fragment serves as an amplification control. Biotin is integrated into PCR amplicon through primer incorporation. AFLP products undergo agarose gel electrophoresis and Southern transfer to a nylon membrane. Amplicons were detected using a streptavidin-enzyme conjugate. Either colorimetric- or chemiluminescent-developed bands are genotyped using locus-specific allele ladders with known VNTR repeat numbers. Using this methodology, we have successfully typed > 500 individuals from three population groups for each locus during data basing and casework.

5/3,AB/68 (Item 68 from file: 155)

DIALOG(R) File 155:MEDLINE(R)

08364005 95207994 PMID: 7900082

Association of a variable number of tandem repeats (VNTR) in glycoprotein Ib alpha and HPA-2 alloantigens.

Simsek S; Bleeker PM; van der Schoot CE; von dem Borne AE

Central Laboratory of The Netherlands Red Cross Blood Transfusion

Service, Amsterdam.

Thrombosis and haemostasis (GERMANY) Nov 1994, 72 (5) p757-61,
ISSN 0340-6245 Journal Code: VQ7

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

The human platelet alloantigen HPA-2(Koa/Kob) system is involved in two clinical syndromes, neonatal alloimmune thrombocytopenia and platelet transfusion refractoriness. We have previously described that the human platelet alloantigens HPA-2a(Kob) and HPA-2b(Koa), are caused by a Thr145Met amino acid polymorphism in the N-terminal globular domain of the human platelet glycoprotein (GP) Ib alpha. In the present study the question was addressed as to whether a genetic association exists between this Thr145Met polymorphism and the recently described variable number of tandem repeat (VNTR) polymorphism in GP Ib alpha. Such an association has already been suggested by serological analysis (Ishida et al., 1991). This VNTR polymorphism results from a 13-amino-acid sequence repeat in the macroglycopeptide region of GP Ib alpha. Therefore, we developed a PCR method to analyze the VNTR region of 106 normal individuals who were also analyzed for the HPA-2 polymorphism. In this method genomic DNA derived from mononuclear cells was purified, the polymorphic region was amplified by PCR and was electrophoresed on agarose gels. Differences in the size of the PCR products made VNTR typing possible. Genotyping for the HPA-2 system was done by allele-specific restriction site analysis of PCR products with the restriction enzyme Sfa NI. The DNA derived from 12 HPA-2(a-b+) subjects, contained only the B variant (with 3 repeats) of the VNTR polymorphism. The D variant (with 1 repeat) was only found in HPA-2a positive individuals. The C variant (with 2 repeats) was found to be strongly associated with HPA-2a. However, two members of a family with a HPA-2(a+b+) genotype were found to be homozygous for the C variant of the VNTR polymorphism. This shows that the C variant can also be associated with HPA-2b. The A variant (with 4 repeats) was not encountered in the population studied. The strong association of HPA-2 and VNTR polymorphism, lying 761 bp apart on the GP Ib alpha gene, indicates linkage disequilibrium.

5/3,AB/69 (Item 69 from file: 155)
DIALOG(R)File 155:MEDLINE(R)

08243858 94234158 PMID: 7909987

Linkage of the VNTR /insulin-gene and type I diabetes mellitus:
increased gene sharing in affected sibling pairs.

Owerbach D; Gabbay KH

Diabetes Research Center, Baylor College of Medicine, Houston.

American journal of human genetics (UNITED STATES) May 1994, 54

(5) p909-12, ISSN 0002-9297 Journal Code: 3IM

Contract/Grant No.: DK-39044, DK, NIDDK

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

Ninety-six multiplex type I diabetic families were typed at the 5' flanking region of the insulin gene by using a PCR assay that better resolves the VNTR into multiple alleles. Affected sibling pairs shared 2, 1, and 0 VNTR alleles--identical by descent--at a frequency of .47, .45, and .08, respectively, a ratio that deviated from the expected 1:2:1 ratio ($P < .001$). These results confirm linkage of the chromosome 11p15.5 region with type I diabetes mellitus susceptibility.

5/3,AB/70 (Item 70 from file: 155)
DIALOG(R)File 155:MEDLINE(R)

08230304 94357582 PMID: 8076940

5/3,AB/72 (Item 72 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

08225804 94295585 PMID: 7912887

Evaluation of 13 short tandem repeat loci for use in personal identification applications.

Hammond HA; Jin L; Zhong Y; Caskey CT; Chakraborty R
Department of Molecular and Human Genetics, Baylor College of Medicine,
Houston, TX 77030.

American journal of human genetics (UNITED STATES) Jul 1994, 55

(1) p175-89, ISSN 0002-9297 Journal Code: 3IM

Comment in Am J Hum Genet. 1995 Apr;56(4) 1005-6

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

Personal identification by using DNA typing methodologies has been an issue in the popular and scientific press for several years. We present a PCR-based DNA-typing method using 13 unlinked short tandem repeat (STR) loci. Validation of the loci and methodology has been performed to meet standards set by the forensic community and the accrediting organization for parentage testing. Extensive statistical analysis has addressed the issues surrounding the presentation of "match" statistics. We have found STR loci to provide a rapid, sensitive, and reliable method of DNA typing for parentage testing, forensic identification, and medical diagnostics. Valid statistical analysis is generally simpler than similar analysis of RFLP-VNTR results and provides powerful statistical evidence of the low frequency of random multilocus genotype matching.

5/3,AB/73 (Item 73 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

08216846 94341750 PMID: 8063275

High sensitive DNA typing approaches for the analysis of forensic evidence: comparison of nested variable number of tandem repeats (VNTR) amplification and a short tandem repeats (STR) polymorphism.

Schmitt C; Schmutzler A; Prinz M; Staak M

Institute of Forensic Medicine, University of Cologne, Germany.

Forensic science international (IRELAND) Jun 3 1994, 66 (2)

p129-41, ISSN 0379-0738 Journal Code: F49

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

The approach of using nested primers for the APO B variable number of tandem repeats (VNTR) increases the sensitivity of the polymerase chain reaction (PCR) to single cell level. Different experiments and a comparison to the short tandem repeats (STR) system VWA were carried out, to determine the applicability of this method to forensic samples. Nested amplification of the Apo B VNTR was affected by a strong tendency towards preferential amplification of the shorter alleles. This phenomenon was observed for DNA quantities as low as 100 pg and impaired, depending on the allele length, the results for mixed samples. As expected, VWA polymorphism showed less preferential amplification. The high sensitivity of both PCR systems is accompanied by an increased susceptibility to contamination. Using artificially contaminated bloodstains, the bloodstain genotype, the contamination or both genotypes could be found on one piece of evidence. Here a single analysis can lead to an incorrect result. Therefore a strategy for obtaining reliable results should consist of multiple stain extractions and the amplification of different stepped dilutions of the DNA solution.

5/3,AB/74 (Item 74 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

08200246 94314290 PMID: 8039774

Analysis of short tandem repeat (STR) HUMVWA in the Spanish population.

Lorente JA; Lorente M; Budowle B; Wilson MR; Villanueva E

Department of Legal Medicine, University of Granada, Spain.

Forensic science international (IRELAND) May 13 1994, 65 (3)

p169-75, ISSN 0379-0738 Journal Code: F49

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

Amplification by polymerase chain reaction (PCR) of variable number of tandem repeat (VNTR) loci and subsequent typing by electrophoresis and silver staining has become a useful tool for identity testing. One viable group of genetic markers amenable to amplification by PCR is the short tandem repeat (STR) loci. A horizontal discontinuous polyacrylamide gel electrophoresis (PAGE) method was used to type the amplified products of the STR HUMVWA. Typing for VWA of 120 unrelated Spanish Caucasians was done. Six alleles were observed with frequencies in the range 0.096-0.242. The genotype distribution meets Hardy-Weinberg expectations ($0.25 < P < 0.50$). The heterozygosity was 73.3% and the discrimination power (DP) 0.94. Simultaneously, in a small sample of families (n = 24) no new mutations could be found.

5/3,AB/75 (Item 75 from file: 155)

DIALOG(R) File 155:MEDLINE(R)

08185250 94290498 PMID: 8019561

A single base mutation in the type II procollagen gene (COL2A1) that converts glycine alpha 1-247 to serine in a family with late-onset spondyloepiphyseal dysplasia.

Ritvaniemi P; Sokolov BP; Williams CJ; Considine E; Yurgenev L; Meerson EM; Ala-Kokko L; Prockop DJ

Collagen Research Unit, University of Oulu, Finland.

Human mutation (UNITED STATES) 1994, 3 (3) p261-7, ISSN

1059-7794 Journal Code: BRD

Contract/Grant No.: AR-39740, AR, NIAMS

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

A search for mutations in the gene for type II procollagen (COL2A1) was carried out in a family with late-onset spondyloepiphyseal dysplasia resulting in short stature, restricted mobility and severe pain in joints, deforming arthritis in the hips, and claudication. Analysis of the HindIII and VNTR polymorphisms at the COL2A1 gene in the family raised the possibility that the gene cosegregated with the disease. Screening for mutations in the COL2A1 gene using PCR-denaturing gradient gel electrophoresis suggested a sequence variation in exon 19 of one allele of the COL2A1 gene in the proband. Direct sequencing of the PCR products for exon 19 revealed a single base mutation that converted the codon of -GGT- for glycine at alpha 1-247 to -AGT-, a codon for serine. The mutant that converted the present in all affected family members, but absent in nonaffected members and in a group of 50 unrelated healthy individuals. It was also absent in 20 unrelated patients with chondrodysplasia and 30 unrelated patients with early-onset osteoarthritis.

5/3,AB/76 (Item 76 from file: 155)

DIALOG(R) File 155:MEDLINE(R)

08156295 94231700 PMID: 8176857

Polymerase chain reaction (PCR)/single-strand conformation polymorphism (SSCP) analysis of the human HLA-DQB regions.

Akiyama K; Yoshii T; Obata F; Kashiwagi N; Ishiyama I

Department of Forensic Medicine, Teikyo University School of Medicine,
Tokyo, Japan.

Nippon hoigaku zasshi (JAPAN) Feb 1994, 48 (1) p38-43, ISSN
0047-1887 Journal Code: KL3

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

Genetic diagnosis of 13 **alleles** of HLA-DQB1 (0501, 0502, 5031, 5032, 0601, 0602, 0604, 0201, 0301, 0302, 3032, 0401 and 0402) from 65 human DNA samples was achieved by applying single-strand conformation polymorphism (SSCP) analysis to DNA fragments amplified by the polymerase chain reaction (PCR) using a convenient primer set for DQB1 (recommendation of the International Histocompatibility Workshop, 1991). Differences between strand images (narrow/distinct or broad/diffuse) from the individual **alleles** and their electrophoretic mobilities are regarded as criteria for confirming the genetic diagnosis of DQB1 **alleles**. This primer set amplifies not only DNA fragments belonging to DQB1, but also to DQB2, and classification of 3 phenotypes (1.1, 1.2 and 1.1/1.2) in the presence of two **alleles** at the latter locus was suggested. Consequently, PCR/SSCP of DNA amplified by this primer enables classification of the phenotypes, at least under our experimental conditions, into 3 x 91 groups. Two advantages of SSCP analysis over VNTR with regard to the use of amplified DNA in forensic practice are described.

5/3,AB/77 (Item 77 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

08153923 94252683 PMID: 8194850

PCR amplification of **alleles** at locus D17S5: detection of new and rare long-length **alleles** by oligoprobing in a survey of Australian populations.

Kijas JM; Fowler JC; Van Daal A

School of Biological Sciences, Flinders University of South Australia, Adelaide.

Human biology; an international record of research (UNITED STATES) Apr 1994, 66 (2) p329-37, ISSN 0018-7143 Journal Code: GDV

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

Alleles of the hypervariable human locus D17S5 were amplified by polymerase chain reaction (PCR) and categorized by length. Unlike other surveys of this locus, the products of amplification were authenticated by Southern analysis using an oligomeric probe directed to part of the 70-base-pair (bp) variable number of tandem repeat (VNTR) region. A small number of unusually long **alleles** were located. In a survey of 201 unrelated Caucasian individuals, 16 **alleles** (size range, 170-1430 bp) and 59 genotypes were observed (heterozygosity, 86.4%; discriminating power, 0.963). In a similar survey of 166 traditional Australian aboriginals, 18 **alleles** (size range, 170-1430 bp) and 46 genotypes were found (heterozygosity, 80.8%; discriminating power, 0.942). The **allele** frequencies differed significantly between these two ethnically distinct populations. Comparisons are made with other anthropologically diverse populations.

5/3,AB/78 (Item 78 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

08118017 94160999 PMID: 8116620

Identification of internal variation in the pseudoautosomal VNTR DXYS17, with nonrandom distribution of the **alleles** on the X and the Y chromosomes.

Decorte R; Wu R; Marynen P; Cassiman JJ
Center for Human Genetics, University of Leuven, Belgium.
American journal of human genetics (UNITED STATES) Mar 1994, 54
(3) p506-15, ISSN 0002-9297 Journal Code: 3IM
Languages: ENGLISH
Document type: Journal Article
Record type: Completed

The PCR technique was used to analyze the DXYS17 locus in the pseudoautosomal region of the X and the Y chromosomes. Analysis on an automated DNA sequencer allowed for sensitive and highly accurate typing of 16 different alleles with a size between 480 and 1,100 bp. Two DXYS17 alleles migrated with the same size on agarose or denaturing polyacrylamide gels but with different mobilities on nondenaturing polyacrylamide gels. Sequence analysis showed that, while an identical number of repeats were present in both alleles, differences in the composition of the units were observed. The origin of these differences was found in the 28- and 33-bp units, which only had a specific repeat pattern at the 5' and 3' ends of the region. The genotype distribution for DXYS17 in a Caucasian population did not deviate from the values expected under Hardy-Weinberg equilibrium. However, the frequency of one allele and one genotype was significantly different between males and females. Segregation analysis showed that this difference was the result of a nonrandom distribution of certain alleles on the sex chromosomes in males.

5/3,AB/79 (Item 79 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

08116801 94157443 PMID: 8113713
Assessment of PCR of the D17S30 locus for forensic identification.
Ivey JN; Atchison BA; Georgalis AM
Victorian Institute of Forensic Pathology, Department of Forensic
Medicine, Monash University, South Melbourne, Australia.
Journal of forensic sciences (UNITED STATES) Jan 1994, 39 (1)
p52-63, ISSN 0022-1198 Journal Code: I5Z
Languages: ENGLISH
Document type: Journal Article
Record type: Completed

PCR analysis of the VNTR locus D17S30 was assessed for its potential use in forensic identification analysis. "Allelic drop-out," the inefficient amplification of some alleles, complicates the interpretation of DNA typing at this locus. PCR conditions were varied in an effort to improve amplification of the alleles at this locus. Such changes included the use of denaturants, formamide and DMSO, to overcome any incomplete denaturation of template strands due to GC content or allele size. Lowering the annealing temperature during the PCR cycle enhanced the amplification of a larger fragment, but this was not related to the D17S30 locus. It appears that the structure of the genome of some individuals rendered PCR amplification inefficient at this locus.

5/3,AB/80 (Item 80 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

08114089 94148512 PMID: 8314300
Patterns of allele losses suggest the existence of five distinct regions of LOH on chromosome 17 in breast cancer.
Kirchweiger R; Zeillinger R; Schneeberger C; Speiser P; Louason G; Theillet C
Erste Frauenklinik, Allgemeines Krankenhaus, Vienna, Austria.
International journal of cancer. Journal international du cancer (UNITED STATES) Jan 15 1994, 56 (2) p193-9, ISSN 0020-7136

Journal Code: GQU

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

Chromosome 17 is a frequent target during breast-cancer formation and progression. It has been shown to be affected by **allele** losses at multiple sites, as well as by DNA amplification. Our aim was to delineate a map of the genetic alterations on chromosome 17 in a given set of breast tumors. To this end we analyzed 151 pairs of tumor and cognate lymphocyte DNAs by Southern blotting with 5 RFLP or **VNTR** probes and by **PCR** at 8 CA repeat polymorphic loci for LOHs. Moreover, we studied DNA amplification of the *evi2*, *erbB2*, *thraI*, *gcsf* and *rara* genes. Data presented here point strongly to the existence of 5 distinct regions of **allele** losses on chromosome 17: 2 on 17p, 3 on 17q. Of the 2 regions on 17p, one involves *tp53* while the second is located more distally toward the telomere. LOH was found in 45.9% and 58.8% respectively. The 3 regions on 17q are located: (i) on the proximal portion of the long arm band q21, corresponding to the *brcaI* region; (ii) in a central region defined by the marker D17S74; (iii) on the distal part of 17q (band q25) characterized by losses of the marker D17S24. Each of these regions presented respectively **allele** losses in 47.5%, 33.3% and 40.8% of the informative tumors. Whereas some tumors presented patterns of LOH consistent with the loss of a complete chromosomal arm or of large portions of the chromosome, a high proportion of the analyzed tumors showed interstitial losses. Amplifications were found in 15% of the tumors and were centered around *erbB2*. (ABSTRACT TRUNCATED AT 250 WORDS)

5/3,AB/81 (Item 81 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

08051721 94085037 PMID: 8261733

TaqI digestion of **PCR** product increases the informativity of St14 **VNTR** for the diagnosis of hemophilia A.

Saksova L; Gecz J; Kadasi L; Ferak V

Institute of Molecular Physiology and Genetics, SAS, Bratislava, Slovakia.

Disease markers (NETHERLANDS) Sep 1993, 11 (2-3) p139-41,
ISSN 0278-0240 Journal Code: DIM

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

Recently, a pair of **PCR** primers have been described that make it possible to amplify a highly polymorphic **VNTR** locus DX552 (St14). **PCR** products range in size from approximately 650 to 3000 bp. Ninety X chromosomes from unrelated Caucasian subjects were investigated. Digestion of the **PCR** products with TaqI revealed the presence of a polymorphic TaqI restriction site within the product 200 bp from the end. This restriction site is present on 60% and absent on 40% of all **alleles**, but the absence is confined solely to the **alleles** 1690 bp (39%) and 2100 bp (1%). Thus, there is a strong allelic association between the most frequent 1690 bp **allele** and the absence of the TaqI restriction site. Determination of this polymorphisms within the St14 **VNTR** region increases the expected heterozygosity at the DXS52 locus from 72% to 80%. This increases the fraction of hemophilia A families where this marker is informative for indirect prenatal diagnosis and carrier identification.

5/3,AB/82 (Item 82 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

08051534 94082505 PMID: 7903130

The prevalence study on restriction fragment length polymorphism analysis

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File 155:MEDLINE(R) 1966-2002/May W1

*File 155: This file will be reloaded. Accession numbers will change.

File 5:Biosis Previews(R) 1969-2002/May W1

(c) 2002 BIOSIS

Set Items Description

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? s vntr

S1 2244 VNTR

? s s1 and allele?

2244 S1

143483 ALLELE?

S2 1243 S1 AND ALLELE?

? s s2 and pcr

1243 S2

190663 PCR

S3 348 S2 AND PCR

? rd

...examined 50 records (50)

...examined 50 records (100)

...examined 50 records (150)

...examined 50 records (200)

...examined 50 records (250)

...examined 50 records (300)

...completed examining records

S4 238 RD (unique items)

? s s4 and py<1998

Processing

238 S4

20864098 PY<1998

S5 155 S4 AND PY<1998

? t s5/3,ab/all

5/3,AB/1 (Item 1 from file: 155)

DIALOG(R) File 155:MEDLINE(R)

10740978 97383761 PMID: 9239742

Prenatal diagnosis of the thalassaemia syndromes by rapid DNA analytical methods.

Kanavakis E; Traeger-Synodinos J; Vrettou C; Maragoudaki E; Tzetzis M; Kattamis C

First Department of Pediatrics, Athens University, St. Sophia's Children's Hospital, Greece.

Molecular human reproduction (ENGLAND) Jun 1997, 3 (6) p523-8,

ISSN 1360-9947 Journal Code: CWO

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

Prenatal diagnostic strategies applied today are based mainly on polymerase chain reaction (PCR) analytical protocols. In Greece a wide range of mutations underlie the thalassaemic haemoglobinopathies, and consequently a variety of PCR-based methods are required to facilitate diagnosis of all potential abnormal genotypes. PCR protocols include those which are relatively simple and others that are technically challenging, but very few have been designed for high

through-put clinical diagnostics. Over a period of 18 months we carried out prenatal diagnosis of 147 pregnancies (150 fetal samples) at risk for a wide range of haemoglobinopathies. This involved the precise characterization of parental genotypes and the subsequent analysis of fetal DNA samples. In this series, 18 different mutations in the alpha- or beta-globin clusters were identified. For the characterization of these mutations, five PCR-based protocols were selected: denaturing gradient gel electrophoresis (DGGE), amplification refractory mutation system (ARMS) PCR, restriction endonuclease analysis of PCR fragments, oligonucleotide hybridization and 'gap' PCR for detection of deletions. To avoid spurious diagnosis due to contamination of fetal samples, two additional methods were used to genotype polymorphic variable nucleotide tandem repeat (VNTR) regions of the genome in parental and fetal samples. Through analysis of the results we assess the advantages and drawbacks of the selected PCR-based protocols for providing routine clinical diagnostics.

5/3,AB/2 (Item 2 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

10296152 98053369 PMID: 9391881

Mutation and haplotype analysis of phenylalanine hydroxylase alleles in classical PKU patients from the Czech Republic: identification of four novel mutations.

Kozak L; Blazkova M; Kuhrova V; Pijackova A; Ruzickova S; St'astna S
Department of Biochemical and Molecular Genetics, Research Institute of Child Health, Brno, Czech Republic.

Journal of medical genetics (ENGLAND) Nov 1997, 34 (11) p893-8
, ISSN 0022-2593 Journal Code: J1F

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

Mutations, haplotypes, and other polymorphic markers in the phenylalanine hydroxylase (PAH) gene were analysed in 133 unrelated Czech families with classical phenylketonuria (PKU). Almost 95% of all mutant alleles were identified, using a combination of PCR and restriction analysis, denaturing gradient gel electrophoresis (DGGE), and sequencing. A total of 30 different mutations, 16 various RFLP/VNTR haplotypes, and four polymorphisms were detected on 266 independent mutant chromosomes. The most common molecular defect observed in the Czech population was R408W (54.9%). Each of the other 29 mutations was present in no more than 5% of alleles and 13 mutations were found in only one PKU allele each (0.4%). Four novel mutations G239A, R270fsdel5bp, A342P, and IVS11nt-8g-->a were identified. In 14 (5.1%) alleles, linked to four different RFLP/VNTR haplotypes, the sequence alterations still remain unknown. Our results confirm that PKU is a heterogeneous disorder at the molecular level. Since there is evidence for the gene flow coming from northern, western, and southern parts of Europe into our Slavic population, it is clear that human migration has been the most important factor in the spread of PKU alleles in Europe.

5/3,AB/3 (Item 3 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

09806735 98277444 PMID: 9615217

Microheterogeneity and AMP-FLP analysis of the 3' flanking interleukin-6 VNTR polymorphism in central Spain.

Arroyo E; Garcia-Sanchez F; Ruiz de la Cuesta JM; Vicario JL
Centro de Transfusion de Madrid, Spain.

Gene geography (ITALY) Apr 1997, 11 (1) p73-9, ISSN 0394-249X
Journal Code: AYL
Languages: ENGLISH

Document type: Journal Article

Record type: Completed

The 3' flanking region of the interleukin-6 gene is polymorphic due to the existence of a hyper-variable region consisting of a number of A + T rich variable repeated DNA sequences (VNTR). We used specific primers to amplify this particular VNTR system by PCR in 222 unrelated normal Spaniards from Madrid, Spain. A model of inheritance comprising of five different allele classes was proposed and frequencies evaluated as follows: B4, 0.635; B3.1, 0.029; B3, 0.270; B2, 0.038; B1, 0.027. Also, examples of inheritance of mendelian microheterogeneity are shown. Heterozygosity index was calculated ($H = 0.5$) and no departure from Hardy-Weinberg equilibrium was observed ($\chi^2 = 0.091$, d. f. 1, $p > 0.75$).

5/3,AB/4 (Item 4 from file: 155)

DIALOG(R) File 155:MEDLINE(R)

09656680 98044820 PMID: 9383563

Infrared fluorescent detection of D1S80 alleles from blood and body fluid collected on IsoCode devices.

Roy R; Middendorf LR

Nebraska State Patrol Criminalistics Laboratory, Lincoln, USA.

BioTechniques (UNITED STATES) Nov 1997, 23 (5) p942-5, ISSN

0736-6205 Journal Code: AN3

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

A genetic locus D1S80 (pMCT 118) containing variable number of tandem repeats (VNTR) has been used extensively in forensic analysis and paternity testing. In the current research, DNA was isolated from blood saliva and nasal secretions collected on two types of IsoCode paper-based devices. The D1S80 locus was amplified using PCR technology, and the alleles were separated by gel electrophoresis and then detected using an infrared (IR) fluorescence automated DNA sequencer. IR-labeled amplification products were generated from human genomic DNA using oligonucleotide primers, which were covalently linked to an infrared fluorescent dye (IRD41) at the 5' end. This system combines IR fluorescence chemistry and laser technology, thus eliminating the need for post-electrophoretic gel handling for the detection of the alleles. Real-time detection after separation of the alleles is valuable for visualization of the data. The VNTR alleles are displayed as familiar autoradiogram-like images, which can also be analyzed by computer. Since DNA is eluted from the IsoCode devices only with sterile distilled water and without time-consuming methods of extraction, amplification can be performed from numerous samples within a short period of time.

5/3,AB/5 (Item 5 from file: 155)

DIALOG(R) File 155:MEDLINE(R)

09486814 94361148 PMID: 7915879

VNTR alleles associated with the alpha-globin locus are haplotype and population related.

Martinson JJ; Boyce AJ; Clegg JB

Institute of Molecular Medicine, University of Oxford, England.

American journal of human genetics (UNITED STATES) Sep 1994, 55

(3) p513-25, ISSN 0002-9297 Journal Code: 3IM

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

The human alpha-globin complex contains several polymorphic restriction-enzyme sites (i.e., RFLPs) linked to form haplotypes and is flanked by two hypervariable VNTR loci, the 5' hypervariable region (HVR) and the more highly polymorphic 3'HVR. Using a combination of RFLP

analysis and PCR, we have characterized the 5'HVR and 3'HVR alleles associated with the alpha-globin haplotypes of 133 chromosomes, and we here show that specific alpha-globin haplotypes are each associated with discrete subsets of the alleles observed at these two VNTR loci. This statistically highly significant association is observed over a region spanning approximately 100 kb. With the exception of closely related haplotypes, different haplotypes do not share identically sized 3'HVR alleles. Earlier studies have shown that alpha-globin haplotype distributions differ between populations; our current findings also reveal extensive population substructure in the repertoire of alpha-globin VNTRs. If similar features are characteristic of other VNTR loci, this will have important implications for forensic and anthropological studies.

5/3,AB/6 (Item 6 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

09474034 97457260 PMID: 9311183

Rapid polymerase chain reaction analysis of St14 (DXS52) VNTR: carrier detection of hemophilia A.

Yang YH; Song KS; Kim IK; Cha DH
Department of Obstetrics and Gynecology, College of Medicine, Yonsei University, Seoul, Korea.

journal of obstetrics and gynaecology research (JAPAN) Aug 1997,
23 (4) p399-406, ISSN 1341-8076 Journal Code: CLG

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

OBJECTIVE: To determine the frequency of St14 VNTR allele in Koreans as a marker of the hemophilia A and to evaluate the efficacy of this marker for carrier detection of hemophilia A METHODS: PCR amplified RFLP analysis of St14 VNTR was done in 312 X-chromosomes of 122 unrelated Korean males and 95 females and the same method was applied to carrier detection in the 2 hemophilia A families. RESULTS: There were 13 alleles of different sizes of St14 VNTR locus appeared in 312 X-chromosomes of unrelated Koreans. For carrier detection of hemophilia A, in the family A, the mother showed 1390/ 1330 bp alleles and the father showed 700 bp allele. The affected son has inherited 1390 bp allele from his mother. The daughter at risk showed 1330/700 bp alleles. In family B, the mother showed 1280/700 bp alleles and the stepfather showed 1390 bp allele. The affected son has inherited 1280 bp allele. The daughter at risk showed 1390/700 bp alleles. And so the daughters of the 2 families were not carriers for hemophilia A. CONCLUSION: PCR analysis of St14 VNTR was a useful tool for carrier detection of hemophilia A.

5/3,AB/7 (Item 7 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

09386365 97330427 PMID: 9186886

Interleukin-1 receptor antagonist allele : is it a genetic link between Henoch-Schonlein nephritis and IgA nephropathy?

Liu ZH; Cheng ZH; Yu YS; Tang Z; Li LS
Research Institute of Nephrology, Jinling Hospital, Nanjing University School of Medicine, People's Republic of China.

Kidney international (UNITED STATES) Jun 1997, 51 (6) p1938-42
, ISSN 0085-2538 Journal Code: KVB

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

Henoch-Schonlein purpura nephritis (HSPN) is a multi-organ systemic vasculitis, which shares many clinical, histological and immunological

features with IgA nephropathy (IgAN). To address whether these two diseases have a common genetic background, the polymorphism of the variable number tandem repeat (VNTR) of IL-1 receptor antagonist (IL-1ra) gene has been analyzed using PCR in patients diagnosed with HSPN (N = 43) and IgAN (N = 97), together with normal controls (N = 98) and patients with acute post-infectious glomerulonephritis (APGN), under the concept that IL-1 might play an important role in mediating pathogenesis of vasculitis and glomerulonephritis. It was found that the allele frequency and carriage rate of the interleukin-1 receptor antagonist allele (IL1RN*2) of the IL-1ra gene increased significantly in HSPN patients as compared to IgAN (P < 0.01), APGN (P < 0.05) and normal subjects (P < 0.01). Interestingly, varied carriage rates of IL1RN*2 were found among various groups of IgAN patients presenting with different clinical manifestations. The carriage rate of IL1RN*2 was significantly higher in patients with recurrent gross hematuria than other groups of IgAN patients (P < 0.01). Furthermore, although the carriage rate of IL1RN*2 was higher in HSPN (46.5%) than average IgAN patients (26.8%; P < 0.01), there was no significant difference in the carriage rate of IL1RN*2 between HSPN and those IgAN patients with recurrent gross hematuria (42.8%; P > 0.05). It suggested that the IL1RN*2 allele might be a genetic marker shared by HSPN and a special group of IgAN patients with recurrent gross hematuria. Our preliminary observation provided a genetic evidence to support the hypothesis that HSPN and certain subgroup of IgAN are closely related diseases. Such an association of the gene polymorphism of IL-1ra between HSPN and IgAN with recurrent gross hematuria might serve as a key to explore their pathogenesis and eventually a specific intervention.

5/3,AB/8 (Item 8 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

09385261 97362896 PMID: 9219360

Infrared fluorescent detection of D1S80 alleles.

Roy R

Nebraska State Patrol Criminalistics Laboratory, Lincoln 68502, USA.

Forensic science international (IRELAND) May 23 1997, 87 (1)

p63-71, ISSN 0379-0738 Journal Code: F49

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

A genetic locus D1S80 containing a variable number of tandem repeats (VNTR) has been used extensively in forensic analysis and paternity testing. In the current research, the D1S80 locus was amplified using polymerase chain reaction (PCR) technology and the alleles detected using a high sensitivity infrared (IR) fluorescence automated DNA sequencer. IR-labeled amplification products were generated using oligonucleotide primers which were covalently linked to an infrared fluorescent dye (IRD41) at the 5'-end. Human genomic DNA (1.0 ng or less) isolated from blood and various simulated forensic samples was successfully amplified using this technology. Allelic bands were detected by incorporation of the IR fluorescent dye into PCR products. Both Long Ranger and polyacrylamide denaturing gels permitted clear resolution of individual alleles that differ by only one repeat unit. In the smaller gels a separation distance of only 15 cm allowed separation of the alleles in less than 2 h from sample loading to visualization. This system combines IR fluorescence chemistry and laser technology thus eliminating the need for post-electrophoretic gel handling for the detection of D1S80 alleles. Real-time detection is valuable for immediate visualization of the data and the alleles are displayed as familiar autoradiogram-like images which can also be analyzed by computer. By loading a 64-lane gel twice it is possible to type at least 120 samples in 1 day using a single gel.

5/3,AB/9 (Item 9 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

09369780 97363498 PMID: 9219791

Significance of interleukin-1beta and interleukin-1 receptor antagonist genetic polymorphism in inflammatory bowel diseases.

Heresbach D; Alizadeh M; Dabadie A; Le Berre N; Colombel JF; Yaouanq J; Bretagne JF; Semana G

Service d'Hepato-Gastro-Enterologie, and Laboratoire d'Epidemiologie et d'Hygiene Hospitaliere, CHRU Pontchaillou, Rennes, France.

American journal of gastroenterology (UNITED STATES) Jul 1997,

92 (7) p1164-9, ISSN 0002-9270 Journal Code: 3HE

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

OBJECTIVE: Genetic susceptibility to inflammatory bowel disease is well recognized. There is also increasing evidence for the activation of the mucosal immune system and the production of inflammatory cytokines, i.e., interleukin (IL)-1ra and IL-1beta in the inflammatory bowel disease. The aim of this study was to analyze the IL-1beta and IL-1ra gene polymorphism and linkage disequilibrium coefficient between the different **alleles** of these genes in patients with Crohn's disease (CD) or ulcerative colitis (UC), according to the severity of the disease. METHODS: Two hundred twenty-eight inflammatory bowel disease patients (87 UC and 141 CD) were included in this study and compared with 113 unrelated controls. The IL-1beta and IL-1ra gene polymorphism was studied after specific amplification of variable regions by PCR. A penta-allelic polymorphism, corresponding to a VNTR region located in intron 2 of the IL-1ra gene, was analyzed, whereas bi-allelic RFLPs displayed by two restriction enzymes (TaqI and AvaI) at position -511 of the IL-1beta gene were analyzed. RESULTS: There was no significant difference of genotype distribution between controls and CD or UC patients. However, surgically treated UC patients were characterized by a higher frequency of genotype IL-1ra 1-2 (39 vs 16%, $p < 0.01$) compared with nonoperated UC patients. Moreover, nonoperated UC patients displayed a lower frequency of IL-1ra **allele** 2 than surgically treated UC patients (14 vs 34%, $p < 0.002$) or controls (14 vs 30%, $p < 0.005$). Furthermore, simultaneous analysis of the IL-1beta and IL-1ra genes that are located in the same region of chromosome 2 revealed that CD patients carrying the IL-1beta **allele** 2 were more often noncarriers of IL-1ra **allele** 2 ($p < 0.005$). Moreover, UC and CD patients were, characterized by a lower frequency of the association of IL-1ra **allele** 2 and IL-1beta **allele** 2 compared with controls (8.3 vs 20.3% and 10.6 vs 20.3%, $p < 0.03$). CONCLUSIONS: IL-1ra and IL-1beta gene polymorphism analysis from a clinical standpoint might help in defining UC prognosis. However, functional studies at both the circulating and mucosal level with stratification on **allele** associations, especially IL-1ra **allele** 2-IL-1beta **allele** 2 subgroups must be realized before therapeutic implications.

5/3,AB/10 (Item 10 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

09348715 97251829 PMID: 9097438

Characterization of the variable-number tandem repeats in vrrA from different Bacillus anthracis isolates.

Jackson PJ; Walthers EA; Kalif AS; Richmond KL; Adair DM; Hill KK; Kuske CR; Andersen GL; Wilson KH; Hugh-Jones M; Keim P

Environmental Molecular Biology Group, Los Alamos National Laboratory, New Mexico 87545, USA. jackson@telomere.lanl.gov

Applied and environmental microbiology (UNITED STATES) Apr 1997,

63 (4) p1400-5, ISSN 0099-2240 Journal Code: 6K6

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

PCR analysis of 198 *Bacillus anthracis* isolates revealed a variable region of DNA sequence differing in length among the isolates. Five polymorphisms differed by the presence of two to six copies of the 12-bp tandem repeat 5'-CAATATCAACAA-3'. This variable-number tandem repeat (**VNTR**) region is located within a larger sequence containing one complete open reading frame that encodes a putative 30-kDa protein. Length variation did not change the reading frame of the encoded protein and only changed the copy number of a 4-amino-acid sequence (QYQQ) from 2 to 6. The structure of the **VNTR** region suggests that these multiple repeats are generated by recombination or polymerase slippage. Protein structures predicted from the reverse-translated DNA sequence suggest that any structural changes in the encoded protein are confined to the region encoded by the **VNTR** sequence. Copy number differences in the **VNTR** region were used to define five different *B. anthracis* **alleles**. Characterization of 198 isolates revealed **allele** frequencies of 6.1, 17.7, 59.6, 5.6, and 11.1% sequentially from shorter to longer **alleles**. The high degree of polymorphism in the **VNTR** region provides a criterion for assigning isolates to five allelic categories. There is a correlation between categories and geographic distribution. Such molecular markers can be used to monitor the epidemiology of anthrax outbreaks in domestic and native herbivore populations.

5/3,AB/11 (Item 11 from file: 155)
DIALOG(R)File 155:MEDLINE(R)

09316286 97290184 PMID: 9144940

The distribution of the vWF **alleles** and genotypes in the Palestinian population.

Khatib H; Ezzughayyar M; Ayesh S

Department of Genetics, Silberman Life Sciences Institute, Hebrew University of Jerusalem, Israel.

Journal of forensic sciences (UNITED STATES) May 1997, 42 (3)

p504-5, ISSN 0022-1198 Journal Code: I5Z

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

Short tandem repeat (STR) loci amplified by **PCR** are known as a useful tool for individual identification and paternity testing. Direct **PCR** amplification from small amounts of whole blood is a rapid and convenient method for population screening for STR and **VNTR** markers. The **allele** frequencies of the vWF locus were determined for 127 unrelated Palestinians. Co-dominant segregation was observed in 20 mother/child pairs. Nine **alleles** were observed, with frequencies ranging from 0.004 to 0.327. Heterozygosity was 79%, and discrimination power was 0.927.

5/3,AB/12 (Item 12 from file: 155)
DIALOG(R)File 155:MEDLINE(R)

09309194 97264332 PMID: 9162705

[Highly polymorphic regions of the genes for apolipoprotein B and angiotensin-converting enzyme in the Udmurt population]

Izuchenie vysokopolimorfnykh uchastkov genov apolipoproteina B i angiotenzin-konvertiruiushchego germenta v populiatsii Udmurtov.

Spitsyn VA; Khort MV; Pogoda TV; Shadrina MI; Slominskii PA; Limborskaia SA

Genetika (RUSSIA) Feb 1997, 33 (2) p269-73, ISSN 0016-6758

Journal Code: FNN

Languages: RUSSIAN

Document type: Journal Article

Record type: Completed

The hypervariable regions of the 3'-end of the apolipoprotein B gene (APOB3'-VNTR) and angiotensin converting enzyme gene (ACE), which had 10-15 **alleles** each, were studied in a sample from the Udmurt population by means of polymerase chain reaction (PCR). From the literature data, the genetic position of Udmurts among 12 groups of Caucasoid, Mongoloid, and Negroid populations was determined. The data obtained by the method of principal components indicated that Udmurts held an isolated position in the northern branch of the Caucasoid race.

5/3,AB/13 (Item 13 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

09254251 97164608 PMID: 9012415

Short **alleles** revealed by PCR demonstrate no heterozygote deficiency at minisatellite loci D1S7, D7S21, and D12S11.

Alonso S; Castro A; Fernandez-Fernandez I; de Pancorbo MM
Department of Cell Biology and Morphological Sciences, School of Medicine and Dentistry, Universidad del Pais Vasco, Vizcaya, Spain.

American journal of human genetics (UNITED STATES) Feb 1997, 60
(2) p417-25, ISSN 0002-9297 Journal Code: 3IM

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

Short **VNTR alleles** that go undetected after conventional Southern blot hybridization may constitute an alternative explanation for the heterozygosity deficiency observed at some minisatellite loci. To examine this hypothesis, we have employed a screening procedure based on PCR amplification of those individuals classified as homozygotes in our databases for the loci D1S7, D7S21, and D12S11. The results obtained indicate that the frequency of these short **alleles** is related to the heterozygosity deficiency observed. For the most polymorphic locus, D1S7, approximately 60% of those individuals previously classified as homozygotes were in fact heterozygotes for a short **allele**. After the inclusion of these new **alleles**, the agreement between observed and expected heterozygosity, along with other statistical tests employed, provide additional evidence for lack of population substructuring. Comparisons of **allele** frequency distributions reveal greater differences between racial groups than between closely related populations.

5/3,AB/14 (Item 14 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

09252183 97142467 PMID: 8988586

Genetic variations at four tetrameric tandem repeat loci in Korean population.

Park SJ; Lee WG; Lee SW; Kim SH; Koo BS; Budowle B; Rho HM

Department of Biology, Inje University, Kimhae, Korea.

Journal of forensic sciences (UNITED STATES) Jan 1997, 42 (1)
p125-9, ISSN 0022-1198 Journal Code: 15Z

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

Allele and genotype frequencies for four tetrameric short tandem repeat (STR) loci, HumFES/FPS, HumFOLP23, HumGABRB15, and HumCYAR04, have been determined by polymerase chain reaction (PCR) amplification and subsequent polyacrylamide gel electrophoresis from approximately 200 genetically unrelated Koreans. This method allows a single base pair resolution and rapid typing with silver staining. The **allele** and genotype distributions satisfy Hardy-Weinberg expectation. Also, these STR loci have proven to be useful for forensic analyses and paternity tests in which the variable number of tandem repeat (**VNTR**) loci have some

limitations.

5/3,AB/15 (Item 15 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

09182279 97165326 PMID: 9013087

Polymorphism of the apolipoprotein A-IV gene and its significance in lipid metabolism and coronary heart disease in a Japanese population.

Bai H; Saku K; Liu R; Oribe Y; Yamamoto K; Arakawa K

Department of Internal Medicine, Fukuoka University School of Medicine, Japan.

European journal of clinical investigation (ENGLAND) Dec 1996,
26 (12) p1115-24, ISSN 0014-2972 Journal Code: EN3

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

Apolipoprotein A-IV (apo A-IV) is involved in the metabolism of both triglycerides and high-density lipoproteins (HDLs). Apo A-IV has been suggested as participating in several stages of reverse cholesterol transport. Uncertainty about the exact biochemical function of apo A-IV has made the use of genetic apo A-IV polymorphism (variants) attractive in evaluating its physiological role. To date, although some reports indicate that DNA polymorphisms at this locus play an important role in the metabolism of lipids and lipoproteins in western (Caucasian) populations, no similar comprehensive analysis has been performed in a distinct Japanese population. Using DNA sequencing and a restriction fragment length polymorphism (RFLP) study with polymerase chain reaction (PCR), the following **allele** frequencies were established: (a) codon -8 (G-->A, non-synonymous) **allele** 2 = 0 (n = 105); (b) codon 9 (A-->G, synonymous) **allele** 2 = 0.388 (n = 152); (c) codon 347 (A-->T, non-synonymous) **allele** 2 = 0 (n = 900); (d) codon 360 (T-->G, non-synonymous) **allele** 2 = 0 (n = 800); (e) **VNTR** exon 3 [(CTGT)3 and (CTGT)4] (CTGT)3 = 0.262 (n = 105); and (f) MspI (newly detected polymorphic site) polymorphism (C C/T GG) within intron 2, **allele** 2 = 0.096 (n = 193). The frequencies of these polymorphisms, except for that of the newly identified MspI site, are completely different from those reported in western populations. Among the 900 subjects examined, we found one ACT (Thr) to ACG (Thr) synonymous mutation at codon 347, which does not change the primary structure of apo A-IV. The apo A-IV **allele** frequency in patients (166 men and 56 women) with angiographically proven coronary heart disease (CHD) was also studied [codon 9 **allele** 2 = 0.329 (n = 217); **VNTR** exon 3 (CTGT)3 = 0.262 (n = 84); MspI within intron 2, **allele** 2 = 0.092 (n = 222)]. Furthermore, we evaluated serum lipid and lipoprotein levels quantitatively in control subjects and Japanese CHD patients. These polymorphisms did not show any consistent and significant association with lipid and lipoprotein parameters. In addition, no gender-specific effects of apo A-IV polymorphisms on lipid parameters adjusted for confounding factors were observed in either CHD patients or control subjects. Our results indicate that the apo A-IV gene is not a major determinant of the risk for CHD in Japanese.

5/3,AB/16 (Item 16 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

09181933 97202096 PMID: 9049623

Allele frequency distributions at seven DNA hypervariable loci in a population sample from Calabria (southern Italy).

Rose G; De Luca M; Falcone E; Spadafora P; Carrieri G; De Benedictis G
Cell Biology Department, University of Calabria, Italy.

Gene geography (ITALY) Aug 1996, 10 (2) p135-45, ISSN
0394-249X Journal Code: AYL

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

Genotype and allele frequencies at seven Variable Number of Tandem Repeats (VNTR) loci currently used for forensic purposes have been estimated in a population sample from Calabria (south Italy). DNA target regions relevant to four microsatellites (THO.1; REN.4; D12S67; DYS19) and three minisatellites (D1S80; 3'APOB; TPO.10) were amplified by Polymerase Chain Reaction (PCR) and analysed by electrophoresis and ethidium bromide or silver staining. For all loci, the observed genotypes were found to be in agreement with those expected by the Hardy-Weinberg equilibrium. Data on allele frequencies were in line with those found in sample groups from northern or central Italy, tested for some of the above polymorphisms.

5/3,AB/17 (Item 17 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

09135709 97068868 PMID: 8912056

Polymorphism analysis of the VNTR locus D17S5 in central Spain.

Arroyo E; Garcia-Sanchez F; Prieto L; Ruiz de la Cuesta JM; Vicario JL
Departamento de Toxicologia y Legislacion Sanitaria, Facultad de Medicina. Universidad Complutense, Madrid, Spain.

International journal of legal medicine (GERMANY) 1996, 109 (2)
p98-9, ISSN 0937-9827 Journal Code: AX1

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

The fragment length polymorphism YNZ22 (D17S5) was analysed for a sample of 207 unrelated individuals living in Madrid (Spanish Caucasians) using PCR methodology and high resolution separation. Hardy-Weinberg expectations (HWE) were calculated after pooling alleles into four groups. No deviations from HWE were detectable using the conventional chi 2-test. The power of discrimination was estimated as 0.96 and the mean paternity exclusion chance as 0.7587. A comparison of the allele frequency distribution with those of other Caucasian groups revealed no major differences.

5/3,AB/18 (Item 18 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

09130569 97084213 PMID: 8930557

Genetic and immunological markers in pouchitis.

Brett PM; Yasuda N; Yiannakou JY; Herbst F; Ellis HJ; Vaughan R; Nicholls RJ; Ciclitira PJ

Gastroenterology Unit, UMDS, London, UK.

European journal of gastroenterology & hepatology (ENGLAND) Oct 1996, 8 (10) p951-5, ISSN 0954-691X Journal Code: B9X

Contract/Grant No.: R01 DK47716, DK, NIDDK

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

OBJECTIVES: Previous studies of the interleukin-1 receptor antagonist (IL-1RN) have found an increased frequency of the associated variable number tandem repeat (VNTR) allele 2 for ulcerative colitis (UC) and further evidence has been reported that this allele is associated with increased severity of several other inflammatory conditions. The HLA type of UC patients has also been implicated in the extent of disease as has the presence of perinuclear anti-neutrophil cytoplasmic antibodies (p-ANCA). We therefore decided to test the hypothesis that the p-ANCA, HLA type or the presence of the IL-1RN allele 2 in patients who received a restorative proctocolectomy for

for the detection of hemophilia A carrier.

Song KS; Lee CH; Chung CS; Lee K; Yang YH; Kim KY
Department of Clinical Pathology, Obstetrics & Gynecology, Yonsei
University College of Medicine, Seoul, Korea.

Yonsei medical journal (KOREA) Sep 1993, 34 (3) p239-42,

ISSN 0513-5796 Journal Code: XRR

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

We have analyzed two (BclI and XbaI) intragenic restriction fragment length polymorphisms (RFLPs) and St14 (DXS52) variable number of tandem repeats (VNTR) by rapid PCR method in 97 unrelated normal subjects. The incidences for positive BclI and XbaI polymorphic sites in the Koreans were 81% and 72%, respectively, which were higher than other ethnic groups but similar to that reported in the Chinese or Japanese, giving the heterozygosity rate of 0.32 and 0.40, respectively. The amplified allele size was 880 bp with no other polymorphism in the analysis of St14 (DXS52) VNTR. This finding should be taken into account in the planning of a prenatal diagnosis program for ethnic Koreans.

5/3,AB/83 (Item 83 from file: 155)

DIALOG(R) File 155:MEDLINE(R)

08008039 93298657 PMID: 8100143

Amplified fragment length polymorphism analysis of the VNTR locus D1S80 in central Spain.

Alonso A; Martin P; Albarran C; Sancho M

Instituto de Toxicologia, Seccion de Biologia, Madrid, Spain.

International journal of legal medicine (GERMANY) 1993, 105 (6)

p311-4, ISSN 0937-9827 Journal Code: AX1

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

The polymorphism of the D1S80 locus has been analyzed in a population sample of 203 unrelated individuals living in Madrid (central Spain) by PCR and subsequent semi-dry discontinuous polyacrylamide gel electrophoresis (Tris-chloride/Tris-glycine buffer system) followed by silver staining. The electrophoretic system described in this study offers high resolution in the separation of the different D1S80 alleles allowing the detection of microvariability around the allele T22 in the spanish population. Twenty different alleles containing 17-40 repeats of the basic 16 bp unit were distinguished. The alleles T18 and T24 were found to be relatively common in Spain, as in other populations, with frequencies of 0.224 and 0.372, respectively. No evidence of significant deviations from Hardy-Weinberg equilibrium was found in these preliminary population data.

5/3,AB/84 (Item 84 from file: 155)

DIALOG(R) File 155:MEDLINE(R)

07976647 94085952 PMID: 8262515

Analysis of 6 VNTR loci by 'multiplex' PCR and automated fluorescent detection.

Tully G; Sullivan KM; Gill P

Central Research and Support Establishment, Home Office Forensic Science, Aldermaston, Reading, Berkshire, UK.

Human genetics (GERMANY) Dec 1993, 92 (6) p554-62, ISSN

0340-6717 Journal Code: GED

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

The polymerase chain reaction was used to amplify six small variable

number of tandem repeat loci in two reactions (D19S20 co-amplifying with D17S5 and D1S80; D17S766 co-amplifying with D16S83 and D17S24). When coupled with fluorescent detection of the products, this provides a rapid, highly discriminating automated test. Preferential amplification of small alleles, leading to 'allelic dropout' was found to occur in D19S20 and D16S83. Population databases are presented for Caucasians and Afro-Caribbeans at loci D19S20, D16S83 and D17S24, and for Asians at D19S20.

5/3,AB/85 (Item 85 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

07962333 94063938 PMID: 8244406

Rapid identification of **VNTR** alleles of the human thyroid peroxidase gene by **PCR**: a study in a population sample from south Italy.

Rose G; De Luca M; Falcone E; Giacchetto C; De Benedictis G
Cell Biology Department, University of Calabria, Italy.
Genomics (UNITED STATES) Sep 1993, 17 (3) p796-8, ISSN
0888-7543 Journal Code: GEN
Languages: ENGLISH
Document type: Journal Article
Record type: Completed

5/3,AB/86 (Item 86 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

07927778 93371823 PMID: 8363830

Limiting detection of an amplification signal for HLA-D region and **VNTR** genes by 32P-PCR.

McDaniel DO; Naftilan J; Barber WH
Department of Surgery/Medicine, University of Alabama, Birmingham 35294.
BioTechniques (UNITED STATES) Jul 1993, 15 (1) p140-5, ISSN
0736-6205 Journal Code: AN3
Contract/Grant No.: AR20614-14, AR, NIAMS; NIAID AI27985, AI, NIAID
Languages: ENGLISH
Document type: Journal Article
Record type: Completed

The limiting detection signal for identification of human genetic markers, such as HLA-D and **VNTR** genes, was determined using DNA isolated from a series of decreasing numbers of lymphocytes carrying the target marker in the polymerase chain reaction (PCR). The PCR procedure was assembled by incorporating 32P-labeled dCTP in the reaction mixture. Primers specific for detection of MHC Class II genes such as HLA-DR1, -DR2, -DRw52 and -DRw53 were utilized when cells were mismatched by one DR type, and primers for the identification of the region of variable number of tandem repeats (VNTRs) were utilized where cells had the same DR types. The 32P-incorporated amplified DNA was analyzed by polyacrylamide gel electrophoresis followed by exposure to x-ray film. The sensitivity of the test varied for different allelic markers as evaluated by amplification of DNA from each set of a mixture of lymphocytes. The target HLA-DR markers were detectable in a cell ratio of as high as 1:100,000, whereas the **VNTR** markers were detectable at a 1:1000 cell ratio. The approach described here offers certain advantages: 1) increased sensitivity, 2) quantitative power, 3) reduced assay time, 4) simplified procedure and 5) less expense. This method provides valuable information for studies involving forensic specimens and marrow engraftment after allogeneic bone marrow transplantation (BMT) that require discrete representation of one allele relative to another in a heterozygous sample where limited quantities of target DNA are available.

5/3,AB/87 (Item 87 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

07888501 93252037 PMID: 8486126

Typing of the 3' hypervariable region of the apolipoprotein B gene: approaches, pitfalls, and applications.

Marz W; Ruzicka V; Fisher E; Russ AP; Schneider W; Gross W
Gustav Embden-Centre of Biological Chemistry, Johann Wolfgang
Goethe-University, Frankfurt/Main, Germany.

Electrophoresis (GERMANY) Mar 1993, 14 (3) p169-73, ISSN
0173-0835 Journal Code: ELE

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

Apolipoprotein B-100 is the principal protein component of lipoproteins with very low, intermediate, and low density. The interaction of apoB-100 with low density lipoprotein (LDL) receptors is responsible for the uptake of LDL into cells. An AT-rich hypervariable region is located adjacent to the 3' end of the apoB gene. It consists of a variable number of tandemly repeated sequences (VNTR). Two approaches were used to analyze this polymorphism. In both, the region harboring the VNTR was amplified with the polymerase chain reaction (PCR). In the first method, fluorescently labeled primers were used in the PCR reactions and products were separated in agarose gels by means of an automated fluorescent fragment analyzer. In the second method, PCR products were analyzed in denaturing polyacrylamide gels and detected with silver staining. Even in the highly sophisticated automated system, agarose gel electrophoresis did not always enable unequivocal assignment of VNTR alleles. In contrast, denaturing polyacrylamide gel electrophoresis made it possible to distinguish the 15 bp differences between the VNTR alleles in a precise and simple manner. The VNTR polymorphism was typed in 234 individuals. Among these were 136 patients with coronary artery disease and 74 healthy controls. Thirteen alleles could be distinguished. The allele containing 49 repeats (VNTR -49) was found in 9.2% of the coronary artery disease patients and in 4.7% of the controls. Thus, the VNTR-49 allele increases relative coronary risk by about twofold. It is concluded that the apoB VNTR polymorphism is a potentially useful genetic marker. Since agarose gel electrophoresis may lead to ambiguous results, we prefer typing by denaturing polyacrylamide gel electrophoresis. (ABSTRACT TRUNCATED AT 250 WORDS)

5/3,AB/88 (Item 88 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

07876734 93229391 PMID: 8471543

PCR-amplification and detection of the human D1S80 VNTR locus. Amplification conditions, population genetics and application in forensic analysis.

Kloosterman AD; Budowle B; Daselaar P
Gerechtelijk Laboratorium van het Ministerie van Justitie, Rijswijk, The Netherlands.

International journal of legal medicine (GERMANY) 1993, 105 (5)
p257-64, ISSN 0937-9827 Journal Code: AX1

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

A series of experiments has been performed to evaluate amplification and typing of the D1S80 VNTR locus. The validation study that has been carried out showed that correct D1S80 typing results can be obtained when a defined amplification protocol and a high-resolution polyacrylamide gel electrophoresis method are used. The use of the Chelex extraction protocol has substantially reduced the processing time. DNA-extraction,

amplification and subsequent typing can be performed in one day. The discrimination power of this locus is 0.94 in a Dutch Caucasian population sample. The system is extremely sensitive: 0.1 ng of genomic DNA gave a correct typing result. The test could also detect the correct genotypes in mixed samples containing DNA from different individuals. Even if the major type was in a 20-fold excess, the minority type could still be amplified and typed correctly. We have found no deviation from Hardy-Weinberg equilibrium in a Dutch Caucasian population sample. Evidence for the somatic stability of this locus was obtained from a set of experiments where we compared DNA-profiles from corresponding blood, semen and saliva samples. The results of this study suggest that in the near future analysis of the D1S80 locus by DNA-amplification can be applied in actual forensic case work.

5/3,AB/89 (Item 89 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

07859047 93194185 PMID: 8449503

Heteroduplex analysis can increase the informativeness of PCR-amplified VNTR markers: application using a marker tightly linked to the COL2A1 gene.

Wilkin DJ; Koprivnikar KE; Cohn DH

Steven Spielberg Pediatric Research Center, Ahmanson Department of Pediatrics, Cedars-Sinai Medical Center, Los Angeles, California 90048.

Genomics (UNITED STATES) Feb 1993, 15 (2) p372-5, ISSN 0888-7543 Journal Code: GEN

Contract/Grant No.: HD22657, HD, NICHD

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

Variable number of tandem repeat (VNTR) polymorphisms provide a high degree of informativeness in linkage studies. Whether performed by standard methods or by polymerase chain reaction (PCR), analysis of these markers involves assessment of the length of each allele.

VNTR alleles usually differ in the number of tandem repeats.

During PCR amplification of a VNTR closely linked to the type II collagen gene (COL2A1), we identified allelic microheterogeneity through the analysis of unique heteroduplexes between amplified strands of the two alleles. In one large pedigree, heteroduplex analysis identified six COL2A1 alleles; standard methods would have identified only three distinct alleles. The identification of these heteroduplexes allowed the determination of the COL2A1 inheritance pattern in the family, which otherwise would have been noninformative.

5/3,AB/90 (Item 90 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

07830162 91169524 PMID: 1672295

Characterization and rapid analysis of the highly polymorphic VNTR locus D4S125 (YNZ32), closely linked to the Huntington disease gene.

Richards B; Horn GT; Merrill JJ; Klinger KW

Department of Genetic Disease Research, Integrated Genetics, Inc., Framingham, Massachusetts.

Genomics (UNITED STATES) Feb 1991, 9 (2) p235-40, ISSN 0888-7543 Journal Code: GEN

Contract/Grant No.: 2-R44-HD-25348-02, HD, NICHD

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

The highly polymorphic VNTR locus pYNZ32 has been more extensively characterized, and its analysis converted to a rapid PCR-based format. DNA sequencing in the areas within and flanking the repeated

segment allowed the design of specific amplification primers. The repeated region of pYNZ32 consists of an imperfectly duplicated 27-bp motif, 16 bases of which are more highly conserved. Allelic products from PCR amplification were resolved into nine different size classes ranging from approximately 1400 to 2200 bp. Additional polymorphism was revealed when the amplified products were analyzed by restriction enzyme digestion. Both the overall size variation and the internal sequence polymorphism were used to determine a heterozygosity value of 86% for YNZ32 in 50 unrelated individuals. The rapid analysis and improved resolution of amplified **alleles** on agarose gels, and the internal variability within YNZ32, increase its diagnostic utility as a **VNTR** and as a linkage marker for the nearby Huntington disease gene.

5/3,AB/91 (Item 91 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

07800270 92250067 PMID: 1349582

A minisatellite and a microsatellite polymorphism within 1.5 kb at the human muscle glycogen phosphorylase (PYGM) locus can be amplified by PCR and have combined informativeness of PIC 0.95.

Iwasaki H; Stewart PW; Dilley WG; Holt MS; Steinbrueck TD; Wells SA; Donis-Keller H

Department of Surgery, Washington University School of Medicine, St. Louis, Missouri 63110.

Genomics (UNITED STATES) May 1992, 13 (1) p7-15, ISSN

0888-7543 Journal Code: GEN

Contract/Grant No.: HG 00469, HG, NHGRI; HG00304, HG, NHGRI

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

We sequenced a genomic clone (pMCMP1), previously reported to detect a **VNTR** polymorphism at the PYGM locus, and found a dinucleotide repeat segment (CA)₁₄(GA)₂₅ and a complex (AT)-repeat-rich segment containing 63 repeats spanning 160 bp. Resolution of PCR-amplified genomic DNA from the (CA)(GA) repeat region on DNA sequencing gels revealed a highly informative polymorphism with **alleles** differing by 2-bp intervals and ranging in size from 156 to 190 bp. Among three racial groups, a total of 18 **alleles** were observed. Fourteen **alleles** were observed in Caucasians (PIC 0.89), 12 **alleles** in American Blacks (PIC 0.89), and 9 **alleles** in Pima Indians (PIC 0.73). PCR amplification of the (AT) repeat region and resolution of the products on DNA sequencing gels revealed a complex variable length polymorphism with **alleles** distributed in size from 367 to 970 bp. Twenty-eight **alleles** were found in American Blacks (PIC 0.94), 6 **alleles** in Pima Indians (PIC 0.70), and 11 **alleles** in Caucasians (PIC 0.71). Comparison of the previously described **VNTR** RFLP **alleles** visualized by Southern hybridization to the PCR products described in this report demonstrated that the polymorphism described in both assays was identical. However, a larger number of **alleles** could be detected from the PCR-amplified products. Combined informativeness, PIC 0.95, for the two polymorphisms was determined from haplotype analysis of 100 Caucasian chromosomes. Therefore, for genotyping purposes, informativeness is maximized from using both polymorphisms.

5/3,AB/92 (Item 92 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

07688251 93145050 PMID: 1490173

Automated DNA profiling by fluorescent labeling of PCR products.

Sullivan KM; Pope S; Gill P; Robertson JM

Central Research and Support Establishment, Forensic Science Service, Reading, Berks, UK.

PCR methods and applications (UNITED STATES) Aug 1992, 2 (1)
p34-40, ISSN 1054-9803 Journal Code: BNV
Languages: ENGLISH
Document type: Journal Article
Record type: Completed

DNA profiling has been automated by the fluorescent tagging of amplified variable number tandem repeat (VNTR) loci. This was achieved by the use of fluorescently labeled primers in the amplification of 10 ng of genomic DNA, coupled with laser detection of the products during electrophoresis. The PCR products are sized by co-electrophoresing a standard size ladder mixed with every sample, thereby eliminating errors in size estimation caused by lane-to-lane differences in migration rate. This increases the precision of VNTR characterization and enables alleles that differ by a single 15-bp repeat to be resolved. The system is capable of high throughput: Twenty-four samples are electrophoresed and analyzed within 6 hr. Also, because four different dyes are available, three different loci can be simultaneously characterized with the fourth dye used for the internal standard. Approximately 100 unrelated British caucasians were analyzed at the loci D1S80, D17S5, and ApoB. The probabilities of two unrelated individuals matching by chance (pM) at these three loci were determined to be 0.065, 0.040, and 0.069, respectively, with a combined pM of 1.8×10^{-4} .

5/3,AB/93 (Item 93 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

07678268 93121052 PMID: 1477658

Preferential PCR amplification of alleles: mechanisms and solutions.

Walsh PS; Erlich HA; Higuchi R
Department of Human Genetics, Roche Molecular Systems, Emeryville, California 94608.

PCR methods and applications (UNITED STATES) May 1992, 1 (4)
p241-50, ISSN 1054-9803 Journal Code: BNV
Languages: ENGLISH
Document type: Journal Article
Record type: Completed

The preferential PCR amplification of one allele relative to another in a heterozygous sample could result in an incorrect or ambiguous genetic typing of that sample. There are several mechanisms that could potentially lead to such preferential PCR amplification. First, preferential amplification can result from significant GC% differences between alleles if the conditions of the reaction (denaturation temperature (Tden), duration at the Tden, salt and co-solvent concentrations, etc.) allow the denaturation of one allele but not the other (differential denaturation). For example, the DQa1.1, -1.2, and -1.3 alleles of the HLA-DQa locus do not amplify at a Tden < 89 degrees C; these same conditions still allow amplification of the DQa2, -3, and -4 alleles. However, no differences in amplification efficiency were found between the different HLA-DQa alleles when the Tden was set at the recommended Tden of 94 degrees C, even after as many as 102 cycles of amplification. Second, for PCR-based genetic typing systems in which the PCR products from different alleles differ in length, preferential amplification of the shorter allelic product can occur. Experiments in which the variable number tandem repeat (VNTR) marker D17S5 (YNZ22) was amplified under various conditions suggest that the smaller allelic products are amplified preferentially when Taq polymerase is limiting. Preferential amplification of VNTR alleles can also occur if the target DNA is sufficiently degraded. Third, when the initial number of genomes sampled is very small, stochastic fluctuation in the number of copies of each allele can result in what appears to be preferential amplification. Finally, less efficient priming of DNA synthesis of one allele versus another can occur because of

mismatches between the primer and the specific allelic template, resulting in preferential amplification of the other allele. General strategies to avoid preferential amplification are discussed.

5/3,AB/94 (Item 94 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

07667412 93052251 PMID: 1427793

Variable number of tandem repeat (VNTR) polymorphism at locus D17S5 (YNZ22) in four ethnically defined human populations.

Deka R; De Croo S; Yu LM; Ferrell RE

Department of Human Genetics, University of Pittsburgh, PA 15261.

Human genetics (GERMANY) Sep-Oct 1992, 90 (1-2) p86-90, ISSN 0340-6717 Journal Code: GED

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

We have analyzed the hypervariable locus D17S5 in four well-defined human populations (Kachari of Northeast India; Dogrib Indian of Canada; New Guinea Highlander of Papua New Guinea; and a relatively homogeneous Caucasian population of North German extraction) using both Southern blot analysis and the polymerase chain reaction (PCR) technique to: (1) compare the efficiency and limitation of Southern blotting versus PCR-based techniques in genotyping variable number of tandem repeat loci, and (2) provide allele frequency data at this locus in these four anthropologically defined populations. Preferential PCR amplification of smaller alleles associated with D17S5 was corrected by lowering the DNA template concentration to 200 ng, and by reducing the extension time to 2 min. A perfect correspondence was observed between the results from Southern blot and PCR analysis in all but one sample. A very large allele, of approximately 24 to 25 repeat units, detected by Southern blotting, could not be amplified by PCR, resulting in an incorrect genotyping rate of less than 0.5%. Considering the grave consequences of mistyping in forensic and paternity testing, it is suggested that heterozygous controls consisting of large and small alleles should be employed in each PCR experiment, and PCR-generated homozygotes should be confirmed by Southern blotting. Significant variation in the number and frequency of alleles at this locus was observed in the four examined populations. A total of 15 different alleles were detected. The average heterozygosity varied from 54% in the Dogrib to 89% in the Kachari. No heterozygote deficiency was observed at this locus in any of the examined populations.

5/3,AB/95 (Item 95 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

07564802 92133611 PMID: 1734717

Amplification and characterization of the retinoblastoma gene VNTR by PCR.

Scharf SJ; Bowcock AM; McClure G; Klitz W; Yandell DW; Erlich HA

Department of Human Genetics, Cetus Corporation, Emeryville, CA 94709.

American journal of human genetics (UNITED STATES) Feb 1992, 50 (2) p371-81, ISSN 0002-9297 Journal Code: 3IM

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

VNTR regions are informative genetic markers for linkage mapping and individual identification. Using PCR, we have developed a procedure for the enzymatic amplification of the VNTR located in the 16th intron of the human retinoblastoma (RB1) gene. We have also prepared a nonisotopically labeled oligonucleotide probe which facilitates detection of the amplification products. In examining 250 individuals from four

different populations, we have detected 11 **alleles** ranging from 650 to 1,800 bp in size. The core repeat is approximately 50 bp in length. On the basis of the observed **allele** frequencies for Caucasian, African-American, and Hispanic populations from the United States and for the Mexican Hispanic population, the heterozygosities have been calculated to be 62%, 75%, 61%, and 50%, respectively. The observed genotype frequencies do not deviate from the values expected under Hardy-Weinberg equilibrium. The effect of varying primer sequences, annealing temperature, and cycle number on the amplification are also discussed. Amplification of this marker may also prove useful for detecting the heterozygosity loss that is associated with tumor formation in retinoblastoma.

5/3,AB/96 (Item 96 from file: 155)
DIALOG(R)File 155:MEDLINE(R)

07526211 91232968 PMID: 1674370
Rapid PCR analysis of the St14 (DXS52) **VNTR**.
Richards B; Heilig R; Oberle I; Storjohann L; Horn GT
Department of Genetic Disease Research, Integrated Genetics, Framingham,
MA 01701.
Nucleic acids research (ENGLAND) Apr 25 1991, 19 (8) p1944,
ISSN 0305-1048 Journal Code: O8L
Contract/Grant No.: 2-R44-HD-25348-02, HD, NICHD
Languages: ENGLISH
Document type: Journal Article
Record type: Completed

5/3,AB/97 (Item 97 from file: 155)
DIALOG(R)File 155:MEDLINE(R)

07521031 91090100 PMID: 1670750
Analysis of the **VNTR** locus D1S80 by the PCR followed by
high-resolution PAGE.
Budowle B; Chakraborty R; Giusti AM; Eisenberg AJ; Allen RC
Forensic Science Research and Training Center, Federal Bureau of
Investigation Academy, Quantico, VA 22135.
American journal of human genetics (UNITED STATES) Jan 1991, 48
(1) p137-44, ISSN 0002-9297 Journal Code: 3IM
Contract/Grant No.: GM41399, GM, NIGMS
Languages: ENGLISH
Document type: Journal Article
Record type: Completed
Allelic data for the D1S80 locus was obtained by using the PCR and
subsequent analysis with a high-resolution, horizontal PAGE technique and
silver staining. Compared with RFLP analysis of **VNTR** loci by Southern
blotting, the approach described in this paper offers certain advantages:
(1) discrete **allele** resolution, (2) minimal measurement error, (3)
correct genotyping of single-band **VNTR** patterns, (4) a nonisotopic
assay, (5) a permanent record of the electrophoretic separation, and (6)
reduced assay time. In a sample of 99 unrelated Caucasians, the D1S80 locus
demonstrated a heterozygosity of 80.8% with 37 phenotypes and 16
alleles. The distribution of genotypes is in agreement with expected
values according to the Hardy-Weinberg equilibrium. Furthermore, the
observed number of **alleles** and the level of heterozygosity, obtained
through the protocol described here, were congruent with each other in
accordance with the expectation of a mutation-drift equilibrium model for a
single, homogeneous, random-mating population. Therefore, the analysis of
D1S80 and similar **VNTR** loci by amplified fragment length polymorphism
(AMP-FLP) may prove useful as models for population genetic issues for
VNTR loci analyzed by RFLP typing via Southern blotting.

5/3,AB/98 (Item 98 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

07518552 91331072 PMID: 1831164

Detection of amplified **VNTR** alleles by direct chemiluminescence: application to the genetic identification of biological samples in forensic cases.

Decorte R; Cassiman JJ

Center for Human Genetics, University of Leuven, Belgium.

EXS (SWITZERLAND) 1991, 58 p371-90, Journal Code: BFZ

Languages: ENGLISH

Document type: Journal Article; Review; Review, Tutorial

Record type: Completed

Minisatellite or variable number of tandem repeat (**VNTR**) regions contain such a high degree of polymorphism that they allow one to construct an individual-specific DNA "fingerprint". Analysis of these sequences by Southern blot however, consumes much DNA and is not applicable to degraded DNA samples often recovered from body-fluid stains found at crime scenes. The polymerase chain reaction (**PCR**) technique may overcome these problems. With oligonucleotide primers flanking the repeat region, amplification of the **VNTR** alleles followed by direct visualization on ethidium bromide-stained agarose gels is possible. In those cases where the **PCR** yield is too low for direct visualization, the product can be blotted to a nylon membrane and hybridized with a labelled internal probe. Alternatively, the **PCR** product can be biotinylated during amplification and visualized by direct chemiluminescence after Southern transfer. The remarkable sensitivity of the **PCR** technique has allowed the detection of genetic polymorphisms in single cells, hair roots and single sperm. A drawback of this very high sensitivity however is that special precautions have to be taken to prevent accidental contamination resulting in erroneous interpretation of the results.

5/3,AB/99 (Item 99 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

07518544 91331059 PMID: 1831156

Population genetics of hypervariable loci: analysis of **PCR** based **VNTR** polymorphism within a population.

Chakraborty R; Fornage M; Gueguen R; Boerwinkle E

Genetics Centers, University of Texas Graduate School of Biomedical Sciences, Houston 77225.

EXS (SWITZERLAND) 1991, 58 p127-43, Journal Code: BFZ

Contract/Grant No.: GM-41399, GM, NIGMS

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

Using a polymerase chain reaction (**PCR**) based method, genotypes at two hypervariable loci (3' to the Apo-B-structural gene and at the ApoC-II gene) were determined by size classification of alleles. Genotype data at the Apo-B locus (Apo-B **VNTR**) were obtained on 240 French Caucasians; the sample size for the ApoC-II **VNTR** was 162. For 160 individuals two-locus genotype data were available. Applications of some recently developed statistical methods to these data indicate that both of these loci are at Hardy-Weinberg equilibrium (HWE) and there is no indication of allelic associations between these two unlinked loci. In addition, the observed numbers of alleles (12 for the Apo-B and 11 for the ApoC-II **VNTR** loci) are also consistent with their respective expectations based on the observed heterozygosities (76.9% for the Apo-B and 85.9% for the ApoC-II loci) suggesting genetic homogeneity of this population-based sample. The multimodal distribution of allele sizes observed for both loci indicate that the production of new alleles at such **VNTR** loci may be caused by more than one molecular mechanism.

The utility of such highly polymorphic loci for human genetic research and forensic applications are discussed in the context of these findings.

5/3,AB/100 (Item 100 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

07445569 91362878 PMID: 1888472

Use of DNA amplification by PCR in the study of the hypervariable region (VNTR) in a forensic medicine setting. Experience with 2 systems: Apo B and YNZ 22]

La utilizzazione dell' amplificazione del DNA mediante PCR nello studio di regioni ipervariabili VNTR in ambito medico-forense. Esperienza con due sistemi: ApoB e YNZ 22.

Giorgetti R; Tagliabracci A; Cingolani M; Ferrara SD

Istituto di Medicina Legale, l'Universita di Ancona.

Bollettino della Societa italiana di biologia sperimentale (ITALY) Jan 1991, 67 (1) p25-30, ISSN 0037-8771 Journal Code: ALS

Languages: ITALIAN

Document type: Journal Article

Record type: Completed

The PCR method has been applied to amplify two Variable-number-Tandem-Repeat (VNTR) sequences. The high polymorphism of these VNTR systems can be usefully applied in medical legal fields such as paternity testing and individual identification. The VNTR systems utilized were: ApoB and YNZ 22. The study was conducted on a three-generation family of thirteen members, whose relationship was previously established using conventional blood systems. The results confirm the Mendelian inheritance of the alleles found and the suitability of the PCR method for forensic purposes.

5/3,AB/101 (Item 101 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

07402756 91293795 PMID: 2066110

DNA diagnosis of hydatidiform mole using the polymerase chain reaction.

Fukuyama R; Takata M; Kudoh J; Sakai K; Tamura S; Shimizu N

Department of Molecular Biology, Keio University School of Medicine, Tokyo, Japan.

Human genetics (GERMANY) Jun 1991, 87 (2) p216-8, ISSN 0340-6717 Journal Code: GED

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

We have used the polymerase chain reaction (PCR) technique for the diagnosis of hydatidiform mole, a trophoblastic disease. For this, we targeted the hypervariable 3' flanking region of the APOB gene (APOB/VNTR) because of its high heterozygosity index (0.61) in the Japanese population. We examined seven clinical cases which were tentatively diagnosed as hydatidiform moles. Five of these revealed DNA segments unique to the paternal APOB allele, allowing us to diagnose a complete mole. The PCR technique for targeting the APOB/VNTR appears useful for early diagnosis of hydatidiform mole.

5/3,AB/102 (Item 102 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

07210452 90196009 PMID: 2316523

Five polymorphic microsatellite VNTRs on the human X chromosome.

Luty JA; Guo Z; Willard HF; Ledbetter DH; Ledbetter S; Litt M

Department of Biochemistry, Oregon Health Sciences University, Portland 97201-3098.

American journal of human genetics (UNITED STATES) Apr 1990, 46
(4) p776-83, ISSN 0002-9297 Journal Code: 3IM
Contract/Grant No.: HD20619, HD, NICHD; R01-GM32500, GM, NIGMS
Languages: ENGLISH
Document type: Journal Article
Record type: Completed

The human genome contains approximately 50,000 copies of an interspersed repeat with the sequence (dT.dG/dA.dC)n, where n = approximately 10-60. We and others have found that several of these repeats have variable lengths in different individuals, with allelic fragments varying in size by multiples of 2 bp. These "microsatellite" variable number of tandem repeats (VNTRs) may be scored by PCR, using unique flanking primers to amplify the repeat-containing regions and resolving the products on DNA sequencing gels. Since few VNTRs have been found on the X chromosome, we screened a flow-sorted X chromosome-specific genomic library for microsatellites. Approximately 25% of the phage clones hybridized to a poly (dT-dG).poly(dA-dC) probe. Of seven X-linked microsatellites present in positive phages, five are polymorphic and three have both eight or more alleles and heterozygosities exceeding 75%. Using PCR to amplify genomic DNAs from hybrid cell panels, we confirmed the X localization of these VNTRs and regionally mapped four of them. The fifth VNTR was regionally mapped by virtue of its tight linkage to DXS87 in Centre du Polymorphisme Humain families. We conclude that whatever factors limit the occurrence of "classical" VNTRs and RFLPs on the X chromosome do not appear to operate in the case of microsatellite VNTRs.

5/3,AB/103 (Item 103 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

07087714 94010897 PMID: 8406461

Molecular characterization of an intragenic minisatellite (VNTR) polymorphism in the human parathyroid hormone-related peptide gene in chromosome region 12p12.1-p11.2.

Pausova Z; Morgan K; Fujiwara TM; Bourdon J; Goltzman D; Hendy GN
Department of Medicine, McGill University, Montreal, Quebec, Canada.
Genomics (UNITED STATES) Jul 1993, 17 (1) p243-4, ISSN
0888-7543 Journal Code: GEN

Languages: ENGLISH
Document type: Journal Article
Record type: Completed

The human parathyroid hormone-related peptide (hPTHrP) gene in chromosome region 12p12.1-p11.2 plays an important role in mammalian development and specifically in skeletogenesis. We have characterized a VNTR polymorphism in the hPTHrP gene that is located in an intron 100-bp downstream of exon VI that encodes a 3' untranslated region. By PCR analysis eight different alleles were identified in a group of 112 unrelated individuals. All eight alleles were sequenced and the repeat unit was identified as the general sequence [G(TA)nC]N, where n = 4 to 11 and N = 3 to 17. This polymorphic sequence-tagged site will be useful for mapping chromosome 12p and will aid in testing for linkage of genetic diseases to the hPTHrP gene.

5/3,AB/104 (Item 104 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

07058720 93321945 PMID: 8330805

Frequency distribution of D1S80 alleles in the German population.

Schnee-Griese J; Blass G; Herrmann S; Schneider HR; Forster R; Bassler G; Pflug W

Landeskriminalamt Baden-Wurttemberg, Stuttgart, Germany.

Forensic science international (SWITZERLAND) May 1993, 59 (2)
p131-6, ISSN 0379-0738 Journal Code: F49

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

The locus D1S80 is a very useful genetic marker system for forensic DNA analysis. It consists of a variable number of tandem repeats (VNTR) and can be analyzed by the polymerase chain reaction (PCR). As accurate data about the distribution of the alleles is one of the most important prerequisites for the application in forensic biology we studied the allele distribution in the German population.

5/3,AB/105 (Item 105 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

07044613 93298665 PMID: 8518202

Improved separation of PCR amplified VNTR alleles by a vertical polyacrylamide gel electrophoresis.

Sajantila A; Lukka M

Department of Human Molecular Genetics, National Public Health Institute, Helsinki, Finland.

International journal of legal medicine (GERMANY) 1993, 105 (6)
p355-9, ISSN 0937-9827 Journal Code: AX1

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

The effect of a stacking gel, the pH and crosslinking agent concentration on the resolution and sharpness of PCR amplified VNTR alleles in a vertical discontinuous polyacrylamide gel electrophoresis system was investigated. The experiments show that the use of a low crosslinking agent concentration, a stacking gel and a wide pH difference between the gel buffer and the electrophoresis buffer at the beginning of the electrophoresis resulted in reduced band width and increasing resolution in silver-stained polyacrylamide gels. The importance of sharp DNA fragments is especially emphasized when analyzing multi-allelic DNA loci, that exhibit alleles differing from only few bp to few dozen bp in length, such as variable number of tandem repeat (VNTR) or short tandem repeat (STR) loci.

5/3,AB/106 (Item 106 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

07036963 93167291 PMID: 8434605

Identification of individuals by analysis of biallelic DNA markers, using PCR and solid-phase minisequencing.

Syvanen AC; Sajantila A; Lukka M

Department of Human Molecular Genetics, National Public Health Institute, Helsinki, Finland.

American journal of human genetics (UNITED STATES) Jan 1993, 52
(1) p46-59, ISSN 0002-9297 Journal Code: 3IM

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

We have developed a new method for forensic identification of individuals, in which a panel of biallelic DNA markers are amplified by the PCR, and the variable nucleotides are detected in the amplified DNA fragments by the solid-phase minisequencing method. A panel of 12 common polymorphic nucleotides located on different chromosomes with reported allele frequencies close to .5 were chosen for the test. The allele frequencies for most of the markers were found to be similar in the Finnish and other Caucasian populations. We also introduce a novel approach for rapid determination of the population frequencies of biallelic markers. By this approach we were able to determine the allele frequencies of the markers in the Finnish population, by quantitative

analysis of three pooled DNA samples representing 3,000 individuals. The power of discrimination and exclusion of the solid-phase minisequencing typing test with 12 markers was similar to that of three VNTR markers that are routinely used in forensic analyses at our institute. The solid-phase minisequencing method was successfully applied to type paternity and forensic case samples. We also show that the quantitative nature of our method allows typing of mixed samples.

5/3,AB/107 (Item 107 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

07030311 93148683 PMID: 8426479

Polymerase chain reaction analysis of allele frequency and loss at the Harvey ras locus in myeloid malignancies.

Arderm JC; Saunders MJ; Hyde K; Lawson R; Yin JA; Lucas GS

University Department of Clinical and Laboratory Haematology, Manchester Royal Infirmary, UK.

Leukemia (ENGLAND) Feb 1993, 7 (2) p258-62, ISSN 0887-6924
Journal Code: LEU

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

The hypothesis that 'rare' variable number tandem repeat (VNTR) alleles of the Harvey ras (Ha-ras) locus are an inherited predisposing factor in myeloid malignancies has been evaluated. We describe an application of the polymerase chain reaction (PCR) which amplifies the VNTR region at the Ha-ras locus and offers a number of advantages over conventional Southern analysis. Ha-ras VNTR genotypes were assigned to 57 normal subjects, 46 patients with acute myeloid leukaemia (AML), 26 with myelodysplastic syndrome (MDS) and 49 with chronic granulocytic leukaemia (CGL). By comparison with previous reports we found significantly higher frequencies of rare alleles (20.2%) in our normal subjects of whom more than 35% had at least one 'rare' allele. The frequencies of rare alleles in the patient groups was not significantly different from the normal group ($\chi^2 = 0.54$, $p = 0.91$). In studies of constitutional and leukaemic DNA from patients with AML, we found that allelic loss at the Ha-ras locus was not a common phenomenon. The improved resolution achievable with PCR compared with Southern analysis was demonstrated by the inability of Southern analysis to resolve six out of 34 PCR heterozygotes. We therefore suggest that previous studies showing linkage between rare Ha-ras alleles and susceptibility to malignancy should be reevaluated using our sensitive PCR technique.

5/3,AB/108 (Item 108 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

07019085 93125581 PMID: 1480191

Assay by polymerase chain reaction (PCR) of multi-allele polymorphisms in the Huntington's disease region of chromosome 4.

Allitto BA; McClatchey AI; Barnes G; Altherr M; Wasmuth J; Frischauf AM; MacDonald ME; Gusella J

Molecular Neurogenetics Laboratory, Massachusetts General Hospital, Boston.

Molecular and cellular probes (ENGLAND) Dec 1992, 6 (6)
p513-20, ISSN 0890-8508 Journal Code: NG9

Contract/Grant No.: HG00169, HG, NHGRI; NS16367, NS, NINDS

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

The Huntington's disease-linked D4S115 marker has been converted from a DNA blot assay to a more sensitive and rapid polymerase chain reaction (

PCR) assay. PCR amplification of a tandem repeat at D4S115 revealed 7 allelic fragments, ranging in size from approximately 610 to 915 bp, differing in their apparent copy number of a approximately 55 bp core repeat. This repeat unit differs strikingly in sequence from the repeat units of other multi-allele markers from chromosome region 4p 16.3, arguing that the VNTR (Variable Number of Tandem Repeats) loci clustered in this region did not arise from a common ancestral sequence. The D4S115 marker can be assayed simultaneously with PCR products from D4S125, D4S95 and D4S43 on a single agarose gel, providing a rapid scan for successful amplification of these difficult-to-assay VNTRs, and for inheritance of the entire candidate Huntington's disease region. This approach should help to increase the speed, informativeness and accuracy of presymptomatic and prenatal linkage testing in this devastating disorder.

5/3,AB/109 (Item 109 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

07012769 92359110 PMID: 1353941

Associations between mutations and a VNTR in the human phenylalanine hydroxylase gene.

Goltsov AA; Eisensmith RC; Konecki DS; Lichter-Konecki U; Woo SL
Howard Hughes Medical Institute, Department of Cell Biology, Baylor College of Medicine, Houston, TX 77030.

American journal of human genetics (UNITED STATES) Sep 1992, 51

(3) p627-36, ISSN 0002-9297 Journal Code: 3IM

Contract/Grant No.: HD-17711, HD, NICHD

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

The HindIII RFLP in the human phenylalanine hydroxylase (PAH) gene is caused by the presence of an AT-rich (70%) minisatellite region. This region contains various multiple of 30-bp tandem repeats and is located 3 kb downstream of the final exon of the gene. PCR-mediated amplification of this region from haplotyped PAH chromosomes indicates that the previously reported 4.0-kb HindIII allele contains three of these repeats, while the 4.4-kb HindIII allele contains 12 of these repeats. The 4.2-kb HindIII fragment can contain six, seven, eight, or nine copies of this repeat. These variations permit more detailed analysis of mutant haplotypes 1, 5, 6, and, possibly, others. Kindred analysis in phenylketonuria families demonstrates Mendelian segregation of these VNTR alleles, as well as associations between these alleles and certain PAH mutations. The R261Q mutation, associated with haplotype 1, is associated almost exclusively with an allele containing eight repeats; the R408W mutation, when occurring on a haplotype 1 background, may also be associated with the eight-repeat VNTR allele. Other PAH mutations associated with haplotype 1, R252W and P281L, do not appear to segregate with specific VNTR alleles. The IVS-10 mutation, when associated with haplotype 6, is associated exclusively with an allele containing seven repeats. The combined use of this VNTR system and the existing RFLP haplotype system will increase the performance of prenatal diagnostic tests based on haplotype analysis. In addition, this VNTR may prove useful in studies concerning the origins and distributions of PAH mutations in different human populations.

5/3,AB/110 (Item 110 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

07004104 93041233 PMID: 1419805

von Willebrand disease family studies: comparison of three methods of analysis of the von Willebrand factor gene polymorphism related to a variable number tandem repeat sequence in intron 40.

Gaucher C; Mercier B; Mazurier C
Laboratoire de Recherche sur l'Hemostase, Centre Regional de Transfusion
Sanguine, Lille, France.

British journal of haematology (ENGLAND) Sep 1992, 82 (1)
p73-80, ISSN 0007-1048 Journal Code: AXC

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

A region with a variable number of tandem ATCT repeats (VNTR) has previously been localized within intron 40 of the von Willebrand factor (vWF) gene. In the present report we describe the use of this polymorphism as a genetic marker to study the inheritance pattern in five families affected with various types of von Willebrand disease (vWD): types I, IIA, IIB, IIC and the newly characterized variant with totally defective FVIII binding. Three means of investigation previously reported, all using polymerase chain reaction (PCR) amplification of this vWF gene region, were compared in terms of informativeness. The two direct single-step procedures analysing only partial sequences of the VNTR region turned out to be less informative (three studies informative out of five) than the third method characterizing the variability of the whole VNTR sequence. This latter approach, based on the analysis of the Alu I restriction pattern of the VNTR region, was informative in all the families investigated, therefore avoiding the need to combine it with other genetic marker studies for efficient gene tracking. In conclusion, this two-step (PCR and digestion) method is the most informative for the characterization of the inheritance of the different subtypes of vWD and for the prenatal diagnosis of its severe forms.

5/3,AB/111 (Item 111 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

06990223 91242331 PMID: 1982729

Family studies in von Willebrand's disease by analysis of restriction fragment length polymorphisms and an intragenic variable number tandem repeat (VNTR) sequence.

Standen GR; Bignell P; Bowen DJ; Peake IR; Bloom AL
Department of Haematology, University of Wales College of Medicine,
Cardiff.

British journal of haematology (ENGLAND) Oct 1990, 76 (2)
p242-9, ISSN 0007-1048 Journal Code: AXC

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

We have previously identified a microsatellite variable number tandem repeat region of the nucleotide sequence ATCT within intron 40 of the von Willebrand factor (vWF) gene. By polymerase chain reaction (PCR) amplification of this region, eight major alleles have been demonstrated in the South Wales population, with an overall heterozygosity rate of 75%. Direct sequencing has shown that the alleles correspond to lengths of between six and 14 ATCT repeats. In the present study we describe the use of this variable repeat sequence and previously reported restriction fragment length polymorphisms (RFLP) to study inheritance patterns in families with type I, IIA and severe type III von Willebrand's disease (vWD). The results confirm that analysis of this precisely localized intragenic locus provides a highly informative marker for gene tracking studies in the major forms of vWD.

5/3,AB/112 (Item 112 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

06930450 93035374 PMID: 1357965

X-linked nephrogenic diabetes insipidus: from the ship Hopewell to RFLP

studies.

Bichet DG; Hendy GN; Lonergan M; Arthus MF; Ligier S; Pausova Z; Kluge R; Zingg H; Saenger P; Oppenheimer E; et al

Departement de Medecine, Hopital du Sacre-Coeur, Universite de Montreal, Quebec, Canada.

American journal of human genetics (UNITED STATES) Nov 1992, 51

(5) p1089-1102, ISSN 0002-9297 Journal Code: 3IM

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

Nephrogenic diabetes insipidus (NDI; designated 304800 in Mendelian Inheritance in Man) is an X-linked disorder with abnormal renal and extrarenal V2 vasopressin receptor responses. The mutant gene has been mapped to Xq28 by analysis of RFLPs, and tight linkage between DXS52 and NDI has been reported. In 1969, Bode and Crawford proposed, under the term "the Hopewell hypothesis," that most cases in North America could be traced to descendants of Ulster Scots who arrived in Nova Scotia in 1761 on the ship Hopewell. They also suggested a link between this family and a large Mormon pedigree. DNA samples obtained from 13 independent affected families, including 42 members of the Hopewell and Mormon pedigrees, were analyzed with probes in the Xq28 region. Genealogical reconstructions were performed. Linkage between NDI and DXS304 (probe U6:2.spl), DXS305 (St35-691), DXS52 (St14-1), DXS15 (DX13), and F8C (F814) showed no recombination in 12 families, with a maximum lod score of 13.5 for DXS52. A recombinant between NDI and DXS304, DXS305, was identified in one family. The haplotype segregating with the disease in the Hopewell pedigree was not shared by other North American families. PCR analysis of the St14 VNTR allowed the distinction of two alleles that were not distinguishable by Southern analysis. Carrier status was predicted in 24 of 26 at-risk females. The Hopewell hypothesis cannot explain the origin of NDI in many of the North American families, since they have no apparent relationship with the Hopewell early settlers, either by haplotype or by genealogical analysis. We confirm the locus homogeneity of the disease by linkage analysis in ethnically diverse families. PCR analysis of the DXS52 VNTR in NDI families is very useful for carrier testing and presymptomatic diagnosis, which can prevent the first manifestations of dehydration.

5/3,AB/113 (Item 113 from file: 155)

DIALOG(R) File 155:MEDLINE(R)

06924827 92238494 PMID: 1349197

Nonsynonymous polymorphic sites in the apolipoprotein (apo) A-IV gene are associated with changes in the concentration of apo B- and apo A-I-containing lipoproteins in a normal population.

von Eckardstein A; Funke H; Schulte M; Erren M; Schulte H; Assmann G
Institut fur Klinische Chemie und Laboratoriumsmedizin,
Zentrallaboratorium, Westfalische Wilhelms-Universitat Munster, Germany.

American journal of human genetics (UNITED STATES) May 1992, 50

(5) p1115-28, ISSN 0002-9297 Journal Code: 3IM

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

The aims of this study were to detect polymorphic sites in the apolipoprotein (apo) A-IV gene, to establish their frequencies, to determine potential haplotypes, and to investigate the role of these polymorphisms in lipid metabolism. A sequencing study of four individuals led to the identification of two synonymous mutations (codons 9 and 54) and three nonsynonymous mutations (Val-8----Met, Gln360----His, and Thr347----Ser) and of a VNTR polymorphism within a series of three or four CTGT repeats in the noncoding region of exon 3. Frequencies of these polymorphisms were determined in 291 students by using naturally occurring (BstEII for the synonymous mutation in codon 54, HinfI for Thr347----Ser,

and Fnu4HI for Gln360----His) or artificially introduced restriction-enzyme cutting sites (BstEII for the synonymous mutation in codon 9 and MamI for Val-8----Met), subsequent to PCR amplification. The four-base deletion/insertion polymorphism and its localization cis or trans to the mutations in codons 347 and 360 were studied by direct sequencing of PCR-amplified DNA from 87 students. Frequencies of the rarer alleles were .007 for apo A-IV-8:Met, .04 for the synonymous mutation in codon 9, .14 for the synonymous mutation in codon 54, .16 for apo A-IV347:Ser, .07 for apo A-IV360:His, and .39 for the four-base of insertion. Apo A-IV360:His in all cases was cis-localized to the (CTGT)3 repeat and apo A-IV347:Thr; and apo A-IV347:Ser was cis-localized to the (CTGT)4 repeat and apo A-IV360:Gln. Four haplotypes formed from these three polymorphic sites were thus found. The apo A-IV347:Ser allele was associated both with significantly lower plasma apo B concentrations in both sexes and with significantly lower LDL-cholesterol concentrations in men. Heterozygous carriers of apo A-IV360:His exhibited significantly higher concentrations of LDL-cholesterol and lower Lp(a) concentrations, compared with apo A-IV360:Gln homozygotes. We could not confirm the previously reported association of apo A-IV360:His with elevated HDL-cholesterol concentrations. In the population, the Val-8----Met polymorphism was not associated with significantly different lipid concentrations, but in a family study the Met-8 allele was associated with lower HDL-cholesterol and higher LDL-cholesterol concentrations. In conclusion, our results indicate an important role of the apo A-IV gene locus in the metabolism of apo B and, to a lesser extent, apo A-I containing lipoproteins.

5/3,AB/114 (Item 114 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

06921712 92134769 PMID: 1346500

Amplification of reproducible allele markers for amplified fragment length polymorphism analysis.

Sajantila A; Puomilahti S; Johnsson V; Ehnholm C
Laboratory for Forensic Serology, National Public Health Institute, Helsinki, Finland.

BioTechniques (UNITED STATES) Jan 1992, 12 (1) p16, 18, 20-2,
ISSN 0736-6205 Journal Code: AN3

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

A procedure for amplification by PCR of reproducible allele markers for amplified fragment length polymorphism (Amp-FLP) analysis is presented. We have prepared markers for the allelic products of the VNTR loci D1S80 (MCT118) and D17S30 (YNZ22) and for the hypervariable VNTR locus close to the 3' end of the apolipoprotein B gene (apoB) by re-amplifying a mixture of PCR products from individuals with known alleles. These allele markers allow precise and discrete determination of the VNTR alleles at these loci using the Amp-FLP technique that should prove suitable in forensic analyses, paternity testing and population studies.

5/3,AB/115 (Item 115 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

06915976 93098246 PMID: 1463014

Characteristics of polymorphism at a VNTR locus 3' to the apolipoprotein B gene in five human populations.

Deka R; Chakraborty R; DeCroo S; Rothhammer F; Barton SA; Ferrell RE
Department of Human Genetics, University of Pittsburgh, PA 15261.

American journal of human genetics (UNITED STATES) Dec 1992, 51
(6) p1325-33, ISSN 0002-9297 Journal Code: 3IM

Contract/Grant No.: GM-41399, GM, NIGMS

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

We have analyzed the **allele** frequency distribution at the hypervariable locus 3' to the apolipoprotein B gene (ApoB 3' **VNTR**) in five well-defined human populations (Kacharis of northeast India, New Guinea Highlanders of Papua New Guinea, Dogrib Indians of Canada, Pehuenche Indians of Chile, and a relatively homogeneous Caucasian population of northern German extraction) by using the **PCR** technique. A total of 12 segregating **alleles** were detected in the pooled sample of 319 individuals. A fairly consistent bimodal pattern of **allele** frequency distribution, apparent in most of these geographically and genetically diverse populations, suggests that the ApoB 3' **VNTR** polymorphism predates the geographic dispersal of ancestral human populations. In spite of the observed high degree of polymorphism at this locus (expected heterozygosity levels 55%-78%), the genotype distributions in all populations (irrespective of their tribal or cosmopolitan nature) conform to their respective Hardy-Weinberg predictions. Furthermore, analysis of the congruence between expected heterozygosity and the observed number of **alleles** reveals that, in general, the **allele** frequency distributions at this locus are in agreement with the predictions of the classical mutation-drift models. The data also show that **alleles** that are shared by all populations have the highest average frequency within populations. These findings demonstrate the potential utility of highly informative hypervariable loci such as the ApoB 3' **VNTR** locus in population genetic research, as well as in forensic medicine and determination of biological relatedness of individuals.

5/3,AB/116 (Item 116 from file: 155)

DIALOG(R) File 155:MEDLINE(R)

06914181 93095314 PMID: 1460793

VNTR polymorphism of the collagen type II, alpha 1 (COL2A1) gene detected by **PCR**.

Katsuura Y; Maeiwa M

Department of Legal Medicine, School of Medicine, University of Tokushima, Japan.

Nippon hoigaku zasshi (JAPAN) Oct 1992, 46 (5) p297-300,

ISSN 0047-1887 Journal Code: KL3

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

The 3' side of the human type II collagen alpha 1 (COL2A1) gene contains a region consisting of a variable number of tandemly repeated short A + T-rich DNA sequences (**VNTR**). We amplified this region accurately by the polymerase chain reaction (**PCR**). Genomic DNA was purified from isolated buffy-coat cells, and thermostable Taq polymerase was used to amplify the target region. The amplification products were directly visualized after polyacrylamide gel electrophoresis. In this way, five **alleles** were distinguished in chromosomes from 33 unrelated Japanese, and named A, B, C, D, and E in decreasing order of length. The relative frequencies of the COL2A1 3' **VNTR alleles** A through E were 0.045, 0.075, 0.469, 0.015, and 0.393, respectively. Co-dominant segregation was observed in two informative families. The COL2A1 3' **VNTR** locus was estimated to have a heterozygosity index of 62% and a polymorphic information content of 0.55.

5/3,AB/117 (Item 117 from file: 155)

DIALOG(R) File 155:MEDLINE(R)

06882880 92408685 PMID: 1528202

Rapid DNA fingerprinting to control for specimen errors in HIV testing by the polymerase chain reaction.

Cassol S; Rudnik J; Salas T; Montpetit M; Pon RT; Sy CT; Read S; Major C; O'Shaughnessy MV

Centre of Excellence for HIV/AIDS, University of British Columbia, Vancouver, Canada.

Molecular and cellular probes (ENGLAND) Aug 1992, 6 (4)

p327-31, ISSN 0890-8508 Journal Code: NG9

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

Variable-number-tandem-repeats (VNTRs) are highly polymorphic and provide informative genetic markers for distinguishing between individuals. We have used PCR amplification of VNTR locus pMCT118 to identify mislabelled specimens submitted for HIV PCR testing. The method is rapid, can be applied to large numbers of samples and eliminates the need for radioactive probes. DNA samples (10 ng) are amplified for 25 cycles using fluorescence-labelled oligonucleotide primers (blue dye). An aliquot of the PCR product is then combined with an internal lane size standard (labelled with a red dye), electrophoresed through a 2% agarose gel on an automated fluorescence DNA fragment analyser and the size and quantity of the fragments determined automatically relative to the internal standard. Fifteen alleles, ranging in size from 398 to 709 bp were readily identified in a random sampling of DNA from 63 unrelated HIV-infected patients. Fragment size was reproducible and corresponded to alleles containing from 16 to 35 repeats of a 16 bp unit. VNTR genotyping will prove useful for resolving discordant results due to specimen mix-up and ensuring that the correct samples have been analyzed.

5/3,AB/118 (Item 118 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

06852605 92121847 PMID: 1685164

Typing of deoxyribonucleic acid (DNA) extracted from compact bone from human remains.

Hochmeister MN; Budowle B; Borer UV; Eggmann U; Comey CT; Dirnhofer R

Department of Forensic Medicine, Institut für Rechtsmedizin, University of Bern, Switzerland.

Journal of forensic sciences (UNITED STATES) Nov 1991, 36 (6)

p1649-61, ISSN 0022-1198 Journal Code: I5Z

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

The application of deoxyribonucleic acid (DNA) typing methods for the potential identification of unknown human remains was investigated. DNA was isolated from compact bone tissue from badly decomposed bodies and from known and unknown human remains, using a decalcification and ion wash procedure. Restriction fragment length polymorphism (RFLP) analysis of variable number of tandem repeats (VNTR) loci yielded results in some cases, but more often the DNA was too degraded to produce RFLP patterns. No RFLP profiles could be obtained from putrefied soft tissues. However, DNA extracted from compact bone tissue of human remains up to eleven years old was successfully amplified using the polymerase chain reaction (PCR) for the VNTR loci D1S80, D17S5, COL2A1, and APO B, as well as the HLA-DQ alpha locus. This is especially significant, since PCR results were obtained from those samples whose DNA had been degraded substantially and had yielded no RFLP patterns. All DNA types determined from the compact bone tissue from decomposed bodies whose identification had been established first by other means (and whose parents or offspring were available for typing) demonstrated mendelian inheritance of the alleles of the loci analyzed. These results suggest that amplification and typing of DNA extracted from compact bone of human remains could be useful in establishing the identity of a person, as well

as in excluding possible false identifications.

5/3,AB/119 (Item 119 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

06846747 91294773 PMID: 1676721

Genetic markers in human bone: I. Deoxyribonucleic acid (DNA) analysis.
Lee HC; Pagliaro EM; Berka KM; Folk NL; Anderson DT; Ruano G; Keith TP;
Phipps P; Herrin GL; Garner DD; et al
Connecticut State Police Forensic Science Laboratory, Meriden.
Journal of forensic sciences (UNITED STATES) Mar 1991, 36 (2)
p320-30, ISSN 0022-1198 Journal Code: ISZ
Languages: ENGLISH
Document type: Journal Article
Record type: Completed

Deoxyribonucleic acid (DNA) was isolated from a number of spongy and compact human bone tissue specimens, and the yield was estimated on a "per milligram of starting tissue" basis. DNA was, in addition, isolated from a number of corresponding blood and bone tissue specimens. Spectrophotofluorometry and ethidium bromide visualization on minigels were used to estimate the quantity and degree of degradation of DNA. The DNA from several blood-bone pairs is shown to give concordant restriction fragment length polymorphism (RFLP) typing results by two different typing protocols with five different single-locus probes. DNA from several additional blood-bone pairs is shown to give concordant results for human leucocyte antigen (HLA)-DQ alpha phenotypes following polymerase chain reaction (PCR) amplification and hybridization to specific allele-specific oligonucleotide (ASO) probes, and for the variable numbers of tandem repeats (VNTR) length polymorphisms 3' to the human apolipoprotein B (APOB) gene following PCR amplification with specific primers and analysis of the products by electrophoresis and ethidium bromide visualization.

5/3,AB/120 (Item 120 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

06812955 92144420 PMID: 1782142

Suitability of PCR methods for forensic investigation. Analysis of the 3'apoB VNTR system in an Italian population sample.

Giorgetti R; Tagliabracci A; Agostini A; Cingolani M; Ferrara SD
Istituto di Medicina Legale, Universita di Ancona, Ospedale Regional, Italy.

International journal of legal medicine (GERMANY) 1991, 104 (5)
p243-6, ISSN 0937-9827 Journal Code: AX1
Languages: ENGLISH

Document type: Journal Article
Record type: Completed

The PCR method has been applied to amplify a Variable Number Tandem Repeat (VNTR) sequence located at the 3' end of the apolipoprotein B (ApoB) gene. The study was conducted on an Italian population sample and in a 3-generation family of 13 members, whose relationships were previously established using conventional blood systems. The allele frequencies found were compared with those reported in the literature. The results also confirmed the Mendelian inheritance of the alleles and the suitability of the PCR method for forensic purposes.

5/3,AB/121 (Item 121 from file: 155)
DIALOG(R) File 155:MEDLINE(R)

06795027 92051364 PMID: 1945869

PCR amplification of large VNTR alleles of D17S5

(YNZ22) locus.

Gecz J

Institute of Molecular Physiology and Genetics, Department of Human Genetics, Bratislava, Czechoslovakia.

Nucleic acids research (ENGLAND) Oct 25 1991, 19 (20) p5806,

ISSN 0305-1048 Journal Code: O8L

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

5/3,AB/122 (Item 122 from file: 155)

DIALOG(R) File 155:MEDLINE(R)

06671671 91037865 PMID: 2230693

Amplification of a variable number of tandem repeats (VNTR) locus (pMCT118) by the polymerase chain reaction (PCR) and its application to forensic science.

Kasai K; Nakamura Y; White R

Department of Human Genetics, University of Utah Health Sciences Center, Salt Lake City.

Journal of forensic sciences (UNITED STATES) Sep 1990, 35 (5)

p1196-200, ISSN 0022-1198 Journal Code: ISZ

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

A genetic locus (D1S58, defined by DNA probe pMCT118) that contains a variable number of tandem repeats (VNTR) has been successfully amplified from a very small amount of genomic deoxyribonucleic acid (DNA) by the polymerase chain reaction (PCR). The DNA sequence of the locus was determined and was found to consist of a 16-base consensus sequence and flanking sequences. Oligonucleotide primers complementary to the flanking sequences were synthesized to serve as primers for amplification of MCT118 by the PCR method. Human genomic DNA isolated from blood (2 ng from each sample) was successfully amplified at the MCT118 locus, and polymorphic bands were detectable by ethidium bromide staining after electrophoresis on polyacrylamide gels. Determination of genotypes at this VNTR locus can now be routinely achieved within 24 h, without the need for Southern blots or radioactive materials. Furthermore, the small size (387 to 723 base pairs) of the DNA fragments produced in the PCR amplification permits good resolution of individual alleles that differ by only one repeat unit. The precise specification of the number of tandem repeats present in each allelic fragment is reproducible from one analysis to another.

5/3,AB/123 (Item 123 from file: 155)

DIALOG(R) File 155:MEDLINE(R)

06669303 91033793 PMID: 2227942

Rapid diagnosis of Miller-Dieker syndrome and isolated lissencephaly sequence by the polymerase chain reaction.

Batanian JR; Ledbetter SA; Wolff RK; Nakamura Y; White R; Dobyns WB; Ledbetter DH

Institute for Molecular Genetics, Baylor College of Medicine, Houston, TX 77030.

Human genetics (GERMANY) Oct 1990, 85 (5) p555-9, ISSN

0340-6717 Journal Code: GED

Contract/Grant No.: HD20619, HD, NICHD

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

Probe YNZ22 (D17S5) is a highly polymorphic, variable number tandem repeat (VNTR) marker previously shown to be deleted in all patients

with the Miller-Dieker syndrome (MDS) but not in patients with isolated lissencephaly sequence (ILS). Primers were constructed to the unique sequence flanking the polymorphic, repetitive region of YNZ22 for amplification by the polymerase chain reaction (PCR). Analysis of 118 normal individuals revealed 12 **alleles** (differing in copy number of a 70-bp repeat unit) ranging in size from 168 to 938 bp. A retrospective study of eight MDS and six ILS patients was consistent with Southern blot analysis in all cases except one. In the latter, a very large **allele** (12 copies of the repeat unit) in a patient and her mother failed to amplify on initial attempts, but was successfully amplified by reducing the concentration of genomic DNA used in the reaction. Prospective studies on two MDS and five ILS patients were successfully performed and confirmed in all cases by Southern blot analysis. From the total sample, restriction fragment length polymorphism (RFLP) analysis was fully informative in four of ten MDS patients and showed a deletion in all four cases. Nine of eleven ILS patients were heterozygous and therefore not deleted for YNZ22. Development of primers for additional polymorphic markers in the Miller-Dieker region will lead to a rapid PCR-based diagnostic approach for all MDS and ILS patients. PCR typing of YNZ22 will also facilitate use of this marker in other applications, including genetic linkage, paternity and forensic studies, and analysis of loss of heterozygosity in tumors.

5/3,AB/124 (Item 124 from file: 155)
DIALOG(R)File 155:MEDLINE(R)

06651418 91000361 PMID: 2206402

Rapid detection of hypervariable regions by the polymerase chain reaction technique.

Decorte R; Cuppens H; Marynen P; Cassiman JJ

Center for Human Genetics, University of Leuven, Belgium.

DNA and cell biology (UNITED STATES) Jul-Aug 1990, 9 (6)

p461-9, ISSN 1044-5498 Journal Code: AF9

Languages: ENGLISH

Document type: Journal Article

Record type: Completed

The polymerase chain reaction (PCR) technique has provided a substantial improvement for the detection and analysis of known genetic polymorphisms. Here, we describe the application of this method for the detection of variable number of tandem repeat (VNTR) sequences. With the use of unique oligonucleotide primers, flanking the repeat sequence, and the thermostable Taq DNA polymerase, the hypervariable regions 3' of the Ha-ras gene, 3' of the apolipoprotein B gene, and 5' to the joining segments of the heavy-chain immunoglobulin gene could be amplified. **Alleles** up to 2,000 bp could be visualized directly on ethidium bromide-stained agarose gels. Larger **alleles** were seen only after traditional Southern blot analysis with an internal probe. The value of this new approach for the detection of VNTRs is illustrated in a case of paternity dispute.

5/3,AB/125 (Item 1 from file: 5)
DIALOG(R)File 5:Biosis Previews(R)
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11756609 BIOSIS NO.: 199900002718

Genetic variations at four tetrameric tandem repeat loci in Korean population.

AUTHOR: Park Su Jeong; Lee Woo Ghil; Lee Seung Whan; Kim Sang Hae; Koo Bon Sung; Budowle Bruce; Rho Hyune Mo(a)

AUTHOR ADDRESS: (a)Dep. Mol. Biol., Seoul Natl. Univ., Seoul 151-742**South Korea

JOURNAL: Journal of Forensic Sciences 42 (1):p125-129 Jan., 1997

ISSN: 0022-1198
DOCUMENT TYPE: Article
RECORD TYPE: Abstract
LANGUAGE: English

ABSTRACT: **Allele** and genotype frequencies for four tetrameric short tandem repeat (STR) loci, HumFES/FPS, HumFOLP23, HumGABRB15, and HumCYAR04, have been determined by polymerase chain reaction (PCR) amplification and subsequent polyacrylamide gel electrophoresis from approximately 200 genetically unrelated Koreans. This method allows a single base pair resolution and rapid typing with silver staining. The **allele** and genotype distributions satisfy Hardy-Weinberg expectation. Also, these STR loci have proven to be useful for forensic analyses and paternity tests in which the variable number of tandem repeat (**VNTR**) loci have some limitations.

1997

5/3,AB/126 (Item 2 from file: 5)
DIALOG(R)File 5:Biosis Previews(R)
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11339844 BIOSIS NO.: 199800121176
Gene diagnosis of hemophilia A by polymorphism of St14 (DXS52) **VNTR**.
AUTHOR: Song Jun; Jin Chunlian; Lin Changkun; et al
AUTHOR ADDRESS: Dep. Med. Genet., Sch. Basic Med. Sci., China Med. Univ.,
Shenyang 110001**China
JOURNAL: Journal of China Medical University 26 (6):p554-556 Dec.,
1997

ISSN: 0258-4646
DOCUMENT TYPE: Article
RECORD TYPE: Abstract
LANGUAGE: Chinese; Non-English
SUMMARY LANGUAGE: Chinese; English

ABSTRACT: We analysed polymorphism of St14 (DXS52) **VNTR** in nomol individuals in Northeastern Region Using **PCR** method and detected 8 **alleles**. The longest fragment was 2.4 kb. The amplified fragments with the highest frequency were 700 bp, which accounted for about 57%. The proportion of detected heterozygosity in females was relatively high. We performed gene diagnosis in 3 families with hemophilia A using this **VNTR** polymorphism. In one family, a female was determined to be a normal individual rather than a carrier. In other 2 families 2 male fetuses were determined as patients.

1997

5/3,AB/127 (Item 3 from file: 5)
DIALOG(R)File 5:Biosis Previews(R)
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11251400 BIOSIS NO.: 199800032732
DNA polymorphism in locus D1S80 in Poland. DNA profiling and detection of new **alleles** by heteroduplex formation between **alleles** of the same size.
AUTHOR: Kwiatkowska Jolanta; Dziechciowska Katarzyna; Lisiecka Dobrawa; Slomski Ryszard(a)
AUTHOR ADDRESS: (a)Inst. Hum. Genetics, Polish Acad. Sci., Strzeszynska 32, 60-479 Poznan**Poland
JOURNAL: Journal of Applied Genetics 38 (3):p335-341 1997
ISSN: 1234-1983
DOCUMENT TYPE: Article

RECORD TYPE: Abstract
LANGUAGE: English

ABSTRACT: We have analysed **allele** distribution at the highly polymorphic variable number of tandem repeats (**VNTR**) locus D1S80 (pMCT118) in the Polish population using the polymerase chain reaction (**PCR**) technique. Characteristics of the D1S80 locus makes it a very useful marker for population genetic research, genetic link-age studies and forensic identification of individuals. During our routine application of the D1S80 marker to paternity testing in several cases of homozygosity detected by polyacrylamide gel electrophoresis, heteroduplex formation for **alleles** 18 and 24 was also observed. Direct sequencing of **PCR** products revealed that **alleles** 18 and 24 of locus D1S80 actually represent a mixture composed of different sequences. Our observations indicate that identification of some 18 and 24 **VNTR alleles** based only on size estimated in electrophoretic analyses could lead to errors in paternity testing and DNA profiling.

1997

5/3,AB/128 (Item 4 from file: 5)
DIALOG(R)File 5:Biosis Previews(R)
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11224577 BIOSIS NO.: 199800005909
Infrared fluorescent detection of D1S80 **alleles** from blood and body fluid collected on IsoCode!T!M devices.
AUTHOR: Roy Reena(a); Middendorf Lyle R
AUTHOR ADDRESS: (a)LI-COR Biotechnol. Div., P.O. Box 4000, Lincoln, NE 68504**USA
JOURNAL: Biotechniques 23 (5):p942-945 Nov., 1997
ISSN: 0736-6205
DOCUMENT TYPE: Article
RECORD TYPE: Abstract
LANGUAGE: English

ABSTRACT: A genetic locus D1S80 (pMCT 118) containing variable number of tandem repeats (**VNTR**) has been used extensively in forensic analysis and paternity testing. In the current research, DNA was isolated from blood, saliva and nasal secretions collected on two types of IsoCode paper-based devices. The D1S80 locus was amplified using **PCR** technology, and the **alleles** were separated by gel electrophoresis and then detected using an infrared (IR) fluorescence automated DNA sequencer IR-labeled amplification products were generated from human genomic DNA using oligonucleotide primers, which were covalently linked to an infrared fluorescent dye (IRD41) at the 5' end. This system combines IR fluorescence chemistry and laser technology, thus eliminating the need for post-electrophoretic gel handling for the detection of the **alleley**. Real-time detection after separation of the **alleles** is valuable for visualization of the data. The **VNTR alleles** are disproved as familiar autoradiogram-like images, which can also be analyzed by computer. Since DNA is eluted from the IsoCode devices only with sterile distilled water and without time-consuming methods of extraction, amplification can be performed from numerous samples within a short period of time.

1997

5/3,AB/129 (Item 5 from file: 5)
DIALOG(R)File 5:Biosis Previews(R)
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11132089 BIOSIS NO.: 199799753234

A study on the hypervariable regions of the apolipoprotein B and angiotensin converting enzyme genes in the Udmurt population.

AUTHOR: Spitsyn V A(a); Khort M V(a); Pogoda T V; Shadrina M I; Slominskii P A; Limborskaya S A

AUTHOR ADDRESS: (a)Res. Cent. Med. Genet., Russ. Acad. Med. Sci., Moscow 115478**Russia

JOURNAL: Genetika 33 (2):p269-273 1997

ISSN: 0016-6758

RECORD TYPE: Abstract

LANGUAGE: Russian; Non-English

SUMMARY LANGUAGE: Russian; English

ABSTRACT: The hypervariable regions of the 3'-end of the apolipoprotein B gene (APOB3'-VNTR) and angiotensin converting enzyme gene (ACE), which had 10-15 alleles each, were studied in a sample from the Udmurt population by means of polymerase chain reaction (PCR). From the literature data, the genetic position of Udmurts among 12 groups of Caucasoid, Mongoloid, and Negroid populations was determined. The data obtained by the method of principal components indicated that Udmurts held an isolated position in the northern branch of the Caucasoid race.

1997

5/3,AB/130 (Item 6 from file: 5)
DIALOG(R)File 5:Biosis Previews(R)
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10989839 BIOSIS NO.: 199799610984

Interleukin-1 receptor antagonist allele: Is it a genetic link between Henoch-Schonlein nephritis and IgA nephropathy?

AUTHOR: Liu Hi-Hong; Cheng Zhao-Hong; Yu Yu-Sheng; Tang Zheng; Li Lei-Shi (a)

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JOURNAL: Kidney International 51 (6):p1938-1942 1997

ISSN: 0085-2538

RECORD TYPE: Abstract

LANGUAGE: English

ABSTRACT: Henoch-Schnlein purpura nephritis (HSPN) is a multi-organ systemic vasculitis, which shares many clinical, histological and immunological features with IgA nephropathy (IgAN). To address whether these two diseases have a common genetic background, the polymorphism of the variable number tandem repeat (VNTR) of IL-1 receptor antagonist (IL-1ra) gene has been analyzed using PCR in patients diagnosed with HSPN (N = 43) and IgAN (N = 97), together with normal controls (N = 98) and patients with acute post-infectious glomerulonephritis (APGN), under the concept that IL-1 might play an important role in mediating pathogenesis of vasculitis and glomerulonephritis. It was found that the allele frequency and carriage rate of the interleukin-1 receptor antagonist allele (IL1RN*2) of the IL-1ra gene increased significantly in HSPN patients as compared to IgAN (P lt 0.01), APGN (P lt 0.05) and normal subjects (P lt 0.01). Interestingly, varied carriage rates of IL1RN*2 were found among various groups of IgAN patients presenting with different clinical manifestations. The carriage rate of IL1RN*2 was significantly higher in patients with recurrent gross hematuria than other groups of IgAN patients (P lt 0.01). Furthermore, although the carriage rate of IL1RN*2 was higher in HSPN (46.5%) than average IgAN patients (26.8%; P lt 0.01), there was no significant difference in the carriage rate of IL1RN*2 between HSPN and those IgAN patients with recurrent gross hematuria.

(42.8% $P < 0.05$). It suggested that the IL1RN*2 **allele** might be a genetic marker shared by HSPN and a special group of IgAN patients with recurrent gross hematuria. Our preliminary observation provided a genetic evidence to support the hypothesis that HSPN and certain subgroup of IgAN are closely related diseases. Such an association of the gene polymorphism of IL-1ra between HSPN and IgAN with recurrent gross hematuria might serve as a key to explore their pathogenesis and eventually a specific intervention.

1997

5/3,AB/131 (Item 7 from file: 5)
DIALOG(R)File 5:Biosis Previews(R)
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10944503 BIOSIS NO.: 199799565648

Evaluation of the automated fluorescent analysis of the H-ras minisatellite after optimized **PCR** amplification in comparison with a standardized Southern blot technique.

AUTHOR: Gosse Sandrine(a); Sauvaigo Sylvie; Daver Alain; Larra Francis; Saavedra Jacqueline; Marchand Joseph; Bernard-Gallon Dominique; Bignon Yves-Jean

AUTHOR ADDRESS: (a)Lab. Radioanalyse, Centre Paul Papin, 2 Rue Moll, 49033 Angers**France

JOURNAL: International Journal of Oncology 10 (4):p735-740 1997

ISSN: 1019-6439

RECORD TYPE: Abstract

LANGUAGE: English

ABSTRACT: Determination of **allele** sizes, loss of heterozygosity or genetic instability at minisatellite **VNTR** loci, are routinely performed by the conventional Southern technique. We have investigated the potential use of automated DNA sequencer for the analysis of the H-ras minisatellite. We report the modifications of amplification parameters and electrophoresis conditions on the sequencer. Seventy-one colorectal carcinomas and the corresponding normal tissues were amplified with fluorescent-labeled primers, analyzed on sequencer, and concurrently controlled by Southern blotting. The results on sequencer showed that a Hydrolink matrix used in non-denaturing conditions and a specific analysis software facilitate a more accurate fragment size calculation.

1997

5/3,AB/132 (Item 8 from file: 5)
DIALOG(R)File 5:Biosis Previews(R)
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10909896 BIOSIS NO.: 199799531041

Rare Hras1 with **VNTR alleles** in Spanish lung and lymphoid cancer patients.

AUTHOR: Pifarre A(a); Sanchez-Céspedes M; Aldea A I; Calvo R; Moreno I; Vaquero M; Monzo M; Rosell R

AUTHOR ADDRESS: (a)Mol. Biol. Lab. Cancer Med. Oncol. Serv., Univ. Hosp. Germans Trias i Pujol, 08916 Badalona**Spain

JOURNAL: Proceedings of the American Association for Cancer Research Annual Meeting 38 (0):p243-244 1997

CONFERENCE/MEETING: Eighty-eighth Annual Meeting of the American Association for Cancer Research San Diego, California, USA April 12-16, 1997

ISSN: 0197-016X

RECORD TYPE: Citation

LANGUAGE: English

1997

5/3,AB/133 (Item 9 from file: 5)
DIALOG(R)File 5:Biosis Previews(R)
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10834567 BIOSIS NO.: 199799455712
Study of the D-17S30 locus polymorphism in Chinese population.
AUTHOR: Han Yu Li Shengbin
AUTHOR ADDRESS: Coll. Forensic Med., Xi'an Med. Univ., Xi'an 710061**China
JOURNAL: Xi'an Yike Daxue Xuebao 17 (4):p425-428 1996
ISSN: 0258-0659
RECORD TYPE: Abstract
LANGUAGE: Chinese; Non-English
SUMMARY LANGUAGE: Chinese; English

ABSTRACT: D17S30 is a highly polymorphistic VNTR Locus. Allelic data for the D17S30 Locus was obtained by using PCR and Subsequent analysis with high resolution, horizontal PAGE technic and silver staining. In a sample of 130 unrelated Chinese Han, heterozygosity of the D17S30 Locus was 82. 63% with 42 genotypes and 13 alleles. The allele frequencies were 0.0078-0.3538. The distribution of genotypes was in agreement with expected values according to the Hardy-Weinberg equilibrium. Pedigree analysis in three families confirmed Mendelian inheritance of the alleles. The results showed that D17S30 Locus was a high polymorphism. The analysis of D17S30 and similar VNTR Loci by amplified fragment length polymorphism (Amp-FLP) may prove useful as models for population genetic issues, human identification and genetic diagnosis. It is a great valuable genetic marker in clinical diagnosis, genetic research, forensic identification and paternity testing.

1996

5/3,AB/134 (Item 10 from file: 5)
DIALOG(R)File 5:Biosis Previews(R)
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10788579 BIOSIS NO.: 199799409724
Short alleles revealed by PCR demonstrate no heterozygote deficiency at minisatellite loci D1S7, D7S1 and D12S11.
AUTHOR: Alonso S; Castro A; Fernandez-Fernandez I; De Pancorbo M M
AUTHOR ADDRESS: Dep. Cell Biol. Morphol. Sci., Sch. Med. Dent., 48940 Leioa, Vizcaya**Spain
JOURNAL: American Journal of Human Genetics 60 (2):p417-425 1997
ISSN: 0002-9297
RECORD TYPE: Abstract
LANGUAGE: English

ABSTRACT: Short VNTR alleles that go undetected after conventional Southern blot hybridization may constitute an alternative explanation for the heterozygosity deficiency observed at some minisatellite loci. To examine this hypothesis, we have employed a screening procedure based on PCR amplification of those individuals classified as homozygotes in our databases for the loci D1S7, D7S21, and D12S11. The results obtained indicate that the frequency of these short alleles is related to the heterozygosity deficiency observed. For the most polymorphic locus, D1S7, apprx 60% of those individuals previously classified as homozygotes were in fact heterozygotes for a short allele. After the inclusion of these new alleles, the agreement between observed and expected heterozygosity, along with other statistical tests employed, provide additional evidence for lack of population substructuring. Comparisons of allele frequency

distributions reveal greater differences between racial groups than between closely related populations.

1997

5/3,AB/135 (Item 11 from file: 5)
DIALOG(R)File 5:BIOSIS Previews(R)
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10743507 BIOSIS NO.: 199799364652
Variable number of tandem repeat polymorphism of the endothelial nitric oxide synthase gene in Japanese population.
AUTHOR: Tomita Masafumi(a); Nohno Tsutomu; Okuyama Toshiko; Kawakami Yasuhiko; Ishikawa Tetsuya; Hidaka Kazuo; Ishizu Hideo
AUTHOR ADDRESS: (a)Dep. Legal Med., Okayama Univ. Med. Sch., Okayama 700** Japan
JOURNAL: Kawasaki Medical Journal 22 (2):p51-56 1996
ISSN: 0385-0234
RECORD TYPE: Abstract
LANGUAGE: English

ABSTRACT: We examined the variable number of tandem repeat (VNTR) polymorphism at intron 4 of the human endogenous nitric oxide synthase (eNOS) gene. The **alleles** were amplified from genomic DNA samples by the polymerase chain reaction (PCR), and analyzed by agarose gel electrophoresis. The PCR products showed one or three reproducible bands and two **alleles** and one extra band could be identified. The nucleotide sequence data revealed that the larger **allele**, **allele 2**, was in good agreement with that of the VNTR region, whereas the smaller one, **allele 1**, lacked one 27-bp core unit. The extra band, which showed the largest molecule, always appeared when heterozygote samples were used as templates, indicating a heteroduplex structure resulting from mismatch of the heterozygous **alleles** during annealing. The **allele** frequencies were determined for 112 unrelated Japanese population. The estimated **allele** frequency was **allele 2**=0.87 and **allele 1**=0.13, and the heterozygosity was 0.226. The genotype frequencies were in good accordance with the Hardy-Weinberg equilibrium.

1996

5/3,AB/136 (Item 12 from file: 5)
DIALOG(R)File 5:BIOSIS Previews(R)
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10530447 BIOSIS NO.: 199699151592
Genetic characterization of hTPO VNTR loci in Korean population.
AUTHOR: Lee Ha Kyu(a); Shin Hye Won
AUTHOR ADDRESS: (a)Dep. Biol., Catholic Univ. Korea, Puchon 422-743**South Korea
JOURNAL: Korean Journal of Genetics 18 (2):p117-123 1996
ISSN: 0254-5934
DOCUMENT TYPE: Article
RECORD TYPE: Abstract
LANGUAGE: Korean; Non-English
SUMMARY LANGUAGE: English

ABSTRACT: VNTR (variable number tandem repeat) loci in the genome are characterized as their copy number of the same repeat unit and are extremely polymorphic having potentially hundreds of **alleles** at a single locus. In this study, **allele** distribution on the hTPO VNTR marker was analyzed by PCR (polymerase chain reaction)

in Korean population (100 individuals). Twenty two alleles were identified in the hTPO VNTR locus and alleles 7 (8.5%), 10 (11.0%), and 15 (15.0%) showed three highest peaks. The allele frequency of hTPO VNTR locus is in the 95% confidence interval. PIC (polymorphism information content) value calculated on hTPO VNTR loci was 0.92. POE (power of exclusion) value used in paternity cases was 0.80. MP (matching probability) used in forensic identification was 0.018. In conclusion, statistical data from hTPO VNTR locus showed this locus is a very useful genetic marker for the linkage study, evolutionary genetics and forensic application.

1996

5/3,AB/137 (Item 13 from file: 5)
DIALOG(R)File 5:Biosis Previews(R)
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10513714 BIOSIS NO.: 199699134859
Development of a deoxyribonucleic acid (DNA) restriction fragment length polymorphism (RFLP) database for Punjabis in East Punjab, India.
AUTHOR: Sovinski Sandra M(a); Baird Lynn S; Budowle Bruce; Caruso Joseph F; Cheema Devinder P S; Duncan George T; Hamby Patricia P; Masibay Arni S; Sharma Vijay K; Tahir Mohammad A
AUTHOR ADDRESS: (a)Indianapolis-Marion County Forensic Serv. Agency, 40 South Alabama St., Indianapolis, IN 46204**USA
JOURNAL: Forensic Science International 79 (3):p187-198 1996
ISSN: 0379-0738
DOCUMENT TYPE: Article
RECORD TYPE: Abstract
LANGUAGE: English

ABSTRACT: In response to continuing interest in obtaining reference deoxyribonucleic acid (DNA) analysis data for previously unstudied population groups, blood samples were collected from Punjabi individuals living in East Punjab, India. This first segment of our research is focused on restriction fragment length polymorphism (RFLP) analysis, with future segments anticipated for various polymerase chain reaction (PCR) based techniques. In this study, the samples were subjected to RFLP analysis using HaeIII, followed by hybridization with variable number tandem repeat (VNTR) probes for loci D2S44, D1S7, D10S28, D4S139, D17S79 and D5S110. The band sizes of the resulting patterns were estimated using an FBI imaging system. The resulting data were subjected to statistical analysis for conformity with Hardy-Weinberg expectations, first for the total population of Punjabis, and additionally for the subgroups of Sikhs and Hindus. The loci are highly polymorphic in all sample populations studied. Except for D5S110, there is no evidence for departure from Hardy-Weinberg equilibrium (HWE) for the VNTR loci in the population groups. In addition, there is little evidence of correlation between the alleles at any of the pairs of loci and no evidence of association across the six loci. Finally, the data suggest that a multiple locus VNTR profile would be rare in the Punjabi or either of its subgroups.

1996

5/3,AB/138 (Item 14 from file: 5)
DIALOG(R)File 5:Biosis Previews(R)
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10272580 BIOSIS NO.: 199698727498
Further genetic variability of the VNTR D1S80 (pMCT118):
Correspondence analysis studies.

AUTHOR: Luis J R(a); Caeiro B
AUTHOR ADDRESS: (a)Dep. Antropoloxia, Facultade Bioloxia, Univ. Santiago,
15706 Santiago Compostela, Galicia**Spain
JOURNAL: American Journal of Human Biology 8 (1):p81-87 1996
ISSN: 1042-0533
DOCUMENT TYPE: Article
RECORD TYPE: Abstract
LANGUAGE: English

ABSTRACT: A population genetic study of the VNTR D1S80 (pMCT118 locus) in 206 individuals from the Galician population in Spain was carried out. PCR amplified DNA were electrophoresed in horizontal polyacrylamide gels and subsequently were visualized by silver staining. Up to 19 alleles in 56 different genotypes were found. This report describes a new allele tentatively named T11 that defines the lower limit of repeats reported for this VNTR. A family study demonstrates autosomal codominant inheritance of this allele. Levels of heterozygosity indexes are about 80%. No significant deviations from Hardy-Weinberg equilibrium were observed, using the allele binning method (P gt 0.3 in all cases). Correspondence analysis shows the usefulness of D1S80 alleles in the genetic profiling of human populations, with the alleles 16, 17, 21, 29, and 31 being of particular interest at different levels of analysis.

1996

5/3,AB/139 (Item 15 from file: 5)
DIALOG(R)File 5:Biosis Previews(R)
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10222742 BIOSIS NO.: 199698677660
Population genetics of Atlantic cod using amplified single locus minisatellite VNTR analysis.
AUTHOR: Galvin P; Sadusky T; McGregor D; Cross T
AUTHOR ADDRESS: Zool. Dep., Univ. Coll. Cork, Lee Maltings, Prospect Row,
Cork**Ireland
JOURNAL: Journal of Fish Biology 47 (SUPPL. A):p200-208 1995
ISSN: 0022-1112
DOCUMENT TYPE: Article
RECORD TYPE: Abstract
LANGUAGE: English

ABSTRACT: Atlantic cod, *Gadus morhua*, have been examined extensively over the last two decades using allozyme electrophoresis. More recently, several populations have been studied using mitochondrial DNA (RFLP and sequence) analysis, together with multilocus minisatellite DNA and also microsatellite DNA analyses. The declining status of cod populations in many areas highlights the need for powerful genetic markers capable of discriminating between cod populations, in order to facilitate the design of effective management strategies. Single locus minisatellite DNA analysis offers a potentially powerful alternative to the already mentioned techniques, by combining the power of detection of hypervariable DNA, with non-radioactive techniques in a cost-effective way. As a preliminary investigation into the feasibility of using this approach, four Atlantic cod samples (North Norway, Irish Sea, Scotian Shelf and Northern Cod) were screened at a single polymerase chain reaction (PCR) amplified minisatellite locus (Mmer-AMP2), using primers designed for the flanking regions of a whiting *Merlangius merlangus* L., minisatellite DNA locus. PCR products separated by agarose electrophoresis and viewed by ethidium bromide fluorescence under UV illumination, consisted of one or two bands per individual (corresponding to homozygotes and heterozygotes, respectively). Twenty-two alleles were resolved in 119 cod, and sample

heterozygosity ranged from 0.76 to 0.90. Samples from opposite sides of the Atlantic showed highly significant differences in allelic composition. The results suggest that single locus minisatellite DNA analysis may be of substantial benefit to furthering our knowledge of the population genetics of Atlantic cod.

1995

5/3,AB/140 (Item 16 from file: 5)
DIALOG(R)File 5:Biosis Previews(R)
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10152579 BIOSIS NO.: 199698607497
Polymorphic microsatellite loci from Atlantic salmon (*Salmo salar*): Genetic differentiation of North American and European populations.
AUTHOR: McConnell Stewart K(a); O'Reilly Patrick; Hamilton Lorraine; Wright Jonathan M; Bentzen Paul
AUTHOR ADDRESS: (a)Marine Gene Probe Lab., Dep. Biol., Dalhousie Univ., Halifax, NS B3H 4J1**Canada
JOURNAL: Canadian Journal of Fisheries and Aquatic Sciences 52 (9):p 1863-1872 1995
ISSN: 0706-652X
DOCUMENT TYPE: Article
RECORD TYPE: Abstract
LANGUAGE: English
SUMMARY LANGUAGE: English; French

ABSTRACT: Atlantic salmon populations show low levels of genetic differentiation relative to other salmonid species, when surveyed by allozymes, and with mitochondrial DNA and nuclear ribosomal DNA markers. Here we report the application of three novel microsatellite VNTR loci to population differentiation in Atlantic salmon. A total of 232 microsatellites, cloned from Atlantic salmon, were classified as perfect, imperfect, and compound repeats. Microsatellite length, as in other teleosts, was significantly larger than published mammalian microsatellites. Primers for PCR amplification of three salmon microsatellites were designed. Allele frequencies, degree of polymorphism, and heterozygosity were estimated for five populations from Nova Scotia, Canada, and from Europe. Nei's genetic distances of 0.02-0.9 were observed among populations. There was a clear discrimination between Canadian and European fish based on unique alleles present at two loci. These Atlantic salmon primers also amplify presumably homologous loci in nine other salmonid species. The polymorphic microsatellites loci reported here demonstrate great potential as genetic markers in population, breeding, and evolutionary studies.

1995

5/3,AB/141 (Item 17 from file: 5)
DIALOG(R)File 5:Biosis Previews(R)
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10061123 BIOSIS NO.: 199598516041
Detection of fragment length polymorphism of the VNTR loci D1S80 and D2S123 by PCR amplification, PAGE and silver staining.
AUTHOR: Nam Hyun Suk; Kim Eunhee; Yoon Wan Hee; Lee Kong-Joo(a)
AUTHOR ADDRESS: (a)Coll. Pharm., Ewha Womans Univ., Seoul 120-750**South Korea
JOURNAL: Journal of Biochemistry and Molecular Biology 28 (4):p359-362 1995
ISSN: 1225-8687
DOCUMENT TYPE: Article

RECORD TYPE: Abstract
LANGUAGE: English

ABSTRACT: The highly polymorphic variable number of tandem repeat (VNTR) loci in the human genome are informative markers for the genetic characterization of individuals in the paternity test and forensic science as well as for the study of human disease. In this study, VNTR loci D1S80 and D2S123 have been amplified by PCR and the amplified length polymorphic alleles were detected with a discontinuous vertical PAGE system and silver staining. For explicit DNA typing, PCR optimization, in which amplification efficiencies are similar over a wide range of allele sizes, non-specific amplifications are minimal, and new longer alleles have high amplification efficiency, has been performed by changing the PCR reaction buffer composition and thermal cycling conditions. It turned out that adding an appropriate amount of Tween 20 and NP40 to the PCR reaction buffer and raising the annealing temperature to 68 degree C in thermal cycling made it possible for optimal VNTR loci amplification. A modified PAGE system for VNTR separation was established. Under these conditions, new longer alleles in the D1S80 locus were discovered and D2S123 pattern changes in colorectal tumors were observed. These technical tips are valuable for detecting various amplified fragment length polymorphisms.

1995

5/3,AB/142 (Item 18 from file: 5)
DIALOG(R)File 5:BIOSIS Previews(R)
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09873383 BIOSIS NO.: 199598328301
Variable number tandem repeat (VNTR) polymorphism at locus D17S30 (YNZ22) in a Croatian population sample.
AUTHOR: Kubat Milovan
AUTHOR ADDRESS: Dep. Forensic Med. Criminology, Sch. Med., Univ. Zagreb, Salata 11, 41000 Zagreb**Croatia
JOURNAL: Croatian Medical Journal 36 (1):p23-26 1995
ISSN: 0353-9504
DOCUMENT TYPE: Article
RECORD TYPE: Abstract
LANGUAGE: English

ABSTRACT: Aim. Population sample of 100 individuals living in northwest Croatia (Zagreb area) was tested for YNZ22 polymorphism using the polymerase chain reaction (PCR). Method. After PCR amplification, the phenotypes were separated by polyacrylamide gel electrophoresis, stained with silver stain and identified by comparison with a molecular weight marker and YNZ22 allelic ladder. Results. Thirteen known alleles were identified in the range of 168-1,008 bp. Another larger allele, temporarily designated 13 and still under investigation, was not found in the studied Croatian population. Alleles, pooled in 4 groups to calculate the Hardy-Weinberg equilibrium, showed good accordance between observed and expected values. The power of discrimination was 0.89 and the mean exclusion chance was 0.65. Conclusion. The allele comparison with previous studies on Caucasians showed no significant difference. The high power of discrimination and chance of exclusion make this system a powerful and useful tool for forensic identification and paternity analysis.

1995

5/3,AB/143 (Item 19 from file: 5)

DIALOG(R)File 5:Biosis Previews(R)
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09765431 BIOSIS NO.: 199598220349

Population structure, stepwise mutations, heterozygote deficiency and their implications in DNA forensics.

AUTHOR: Li Jin; Chakraborty Ranajit(a)

AUTHOR ADDRESS: (a)Cent. Demographic Population Genetics, Graduate Sch.
Biomedical Sci., Univ. Texas at Houston Hea**USA

JOURNAL: Heredity 74 (3):p274-285 1995

ISSN: 0018-067X

DOCUMENT TYPE: Article

RECORD TYPE: Abstract

LANGUAGE: English

ABSTRACT: In a substructured population the overall heterozygote deficiency can be predicted from the number of subpopulations (s), their time of divergence (t), and the nature of the mutations. At present the true mutational mechanisms at the hypervariable DNA loci are not known. However, the two existing mutation models (the infinite allele model (IAM) and the stepwise mutation model (SMM)) provide some guides to predictions from which the possible effect of population substructuring may be evaluated, assuming that the subpopulations do not exchange any genes among them during evolution. The theory predicts that the loci with larger mutation rate, and consequently showing greater heterozygosity within subpopulations, should exhibit a smaller proportional heterozygote deficiency (G-ST) and, hence, the effects of population substructuring should be minimal at the hypervariable DNA loci (an order of magnitude smaller than that at the blood group and protein loci). Applications of this theory to data on six Variable Number of Tandem Repeat (VNTR) loci and five short tandem repeat (STR) loci in the major cosmopolitan populations of the USA show that while the VNTR loci often exhibit a large significant heterozygote deficiency, the STR loci do not show a similar tendency. This discordant finding may be ascribed to the limitations, coalescence and nondetectability of alleles associated with the restriction fragment length polymorphism (RFLP) analysis through which the VNTR loci are scored. Such limitations do not apply to the polymerase chain reaction (PCR) method, through which the STR loci are scored. The implications of these results are discussed in the context of the forensic use of DNA typing data.

1995

5/3,AB/144 (Item 20 from file: 5)
DIALOG(R)File 5:Biosis Previews(R)
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09684416 BIOSIS NO.: 199598139334

Inheritance in turnip of variable-number tandem-repeat genetic markers revealed with synthetic repetitive DNA probes.

AUTHOR: Rogstad S H

AUTHOR ADDRESS: Dep. Biol. Sci. ML6, Univ. Cincinnati, Cincinnati, OH
45221-0006**USA

JOURNAL: Theoretical and Applied Genetics 89 (7-8):p824-830 1994

ISSN: 0040-5752

DOCUMENT TYPE: Article

RECORD TYPE: Abstract

LANGUAGE: English

ABSTRACT: Oligomers (16-26 mers) composed of short, tandemly repeated DNA sequences (3-10 bases) were used individually with their complementary oligomer in separate polymerase chain reactions (PCRs) that extended the number of repeats to make 15 different PCR synthetic tandem-repeat

(STR) probes. These PCR-STR probes were used to examine the inheritance of variable-number tandem-repeat (VNTR) genetic markers from two parent plants of turnip (*Brassica rapa* L.) to 20 offspring. Following HinfI digestion and PCR-STR probing of Southern blots, interpretable variable parental and offspring band profiles were found with 9 of the 15 probes used. Each of these nine probes produced a unique set of fragments, and no cases of different probes revealing the same fragment were detected. Seventy-nine parental fragments were found and, of these, 65% (51) appeared to be heterozygous in one or both parents, with 52% (41) appearing to be heterozygous in one of the parents exclusively. That these fragments are transmitted as though heterozygous in the parents implies that they are derived from the nuclear complement of the genome. Chi-square analyses of the transmission of markers are, in general, consistent with Mendelian expectations, although three non-parental bands were found accounting for approximately 0.5% of these transmitted bands. For the fragments heterozygous in one of the parents exclusively, seven alleles exhibited complete linkage in three groups, 12 alleles were incompletely linked in six groups, and four allelic groups involving 11 alleles were identified. PCR-STR probes are relatively rapid to generate and apply (no cloning, clone screening, or sequencing steps are required), and have been shown to reveal VNTR genetic markers in a wide variety of plant species. These results add to the list of studies showing that VNTR genetic markers (and in this case, markers revealed by PCR-STR probes) are transmitted for the greater part in a Mendelian fashion.

1994

5/3,AB/145 (Item 21 from file: 5)
 DIALOG(R)File 5:BIOSIS Previews(R)
 (c) 2002 BIOSIS. All rts. reserv.

09684383 BIOSIS NO.: 199598139301
 An RNA-splicing mutation (G+5IVS20) in the type II collagen gene (COL2A1) in a family with spondyloepiphyseal dysplasia congenita.
 AUTHOR: Tiller George E(a); Weis Mary Ann; Polumbo Paula A; Gruber Helen E; Rimoin David L; Cohn Daniel H; Eyre David R
 AUTHOR ADDRESS: (a)Vanderbilt Univ. Med. Center, Div. Genetics, DD2205 MCN, Nashville, TN 37232-2578**USA
 JOURNAL: American Journal of Human Genetics 56 (2):p388-395 1995
 ISSN: 0002-9297
 DOCUMENT TYPE: Article
 RECORD TYPE: Abstract
 LANGUAGE: English

ABSTRACT: Defects in type II collagen have been demonstrated in a phenotypic continuum of chondrodysplasias that includes achondrogenesis II, hypochondrogenesis, spondyloepiphyseal dysplasia congenita (SEDC), Kniest dysplasia, and Stickler syndrome. We have determined that cartilage from a terminated fetus with an inherited form of SEDC contained both normal alpha-1(II) collagen chains and chains that lacked amino acids 256-273 of the triple-helical domain. PCR amplification of this region of COL2A1, from genomic DNA, yielded products of normal size, while amplification of cDNA yielded a normal sized species and a shorter fragment missing exon 20. Sequence analysis of genomic DNA from the fetus revealed a G to T transversion at position +5 of intron 20; the affected father was also heterozygous for the mutation. Allele-specific PCR and heteroduplex analysis of a VNTR in COL2A1 independently confirmed the unaffected status of a fetus in a subsequent pregnancy. Thermodynamic calculations suggest that the mutation prevents normal splicing of exon 20 by interfering with binding of U-1 small-nuclear RNA to pre-mRNA, thus leading to skipping of exon 20 in transcripts from the mutant allele. Electron micrographs of

diseased cartilage showed intracellular inclusion bodies, which were stained by an antibody to alpha-1(II) procollagen. Our findings support the hypothesis that cc-chain length alterations that preserve the Gly-X-Y repeat motif of the triple helix result in partial intracellular retention of alpha-1(II) procollagen and produce mild to moderate chondrodysplasia phenotypes.

1995

5/3,AB/146 (Item 22 from file: 5)
DIALOG(R)File 5:Biosis Previews(R)
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09642882 BIOSIS NO.: 199598097800

Determination of the frequency of allelic variants of the gene for von Willebrand factor on the basis of polymorphism of a number of tandem repeats in intron 40.

AUTHOR: Misyurin A V; Surin V L; Solov'ev G Ya

AUTHOR ADDRESS: Hematol. Sci. Cent., Russ. Acad. Med. Sci., Moscow 125167
**Russia

JOURNAL: Genetika 30 (5):p713-717 1994

ISSN: 0016-6758

DOCUMENT TYPE: Article

RECORD TYPE: Abstract

LANGUAGE: Russian; Non-English

SUMMARY LANGUAGE: Russian; English

ABSTRACT: The frequencies of allelic variants of the human von Willebrand factor gene (vWF) in a heterogeneous Moscow population were estimated on the basis of the variable number of tandem repeats (vWF.VNTR) in intron 40 using the polymerase chain reaction (PCR). Eighty-nine DNA samples from unrelated individuals were analyzed. The heterozygosity index of polymorphism in the studied population (0.60) was lower than in the population of Wales (0.75). Two previously unknown variants containing 5 and 10 tandem repeats (vWF.VNTR5 and vWF.VNTR9) were found. The phenomenon of "tripling" was observed, it is expressed in some individual cases by the presence of three fragments with different lengths, as observed in the results of (PCR) with the used system of primers. This is linked with the presence of special alleles in these individuals, which are marked by two different fragments rather than one.

1994

5/3,AB/147 (Item 23 from file: 5)
DIALOG(R)File 5:Biosis Previews(R)
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09530775 BIOSIS NO.: 199497539145

VNTR allele distribution in Korean population: Analysis by

PCR and high-resolution agarose gel electrophoresis.

AUTHOR: Chung Y-B; Na W-J

AUTHOR ADDRESS: Inst. Molecular Biol., Paik Hosp., Seoul**South Korea

JOURNAL: American Journal of Human Genetics 55 (3 SUPPL.):pA332 1994

CONFERENCE/MEETING: 44th Annual Meeting of the American Society of Human Genetics Montreal, Quebec, Canada October 18-22, 1994

ISSN: 0002-9297

RECORD TYPE: Citation

LANGUAGE: English

1994

5/3,AB/148 (Item 24 from file: 5)
DIALOG(R)File 5:Biosis Previews(R)
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09275402 BIOSIS NO.: 199497283772

VNTR polymorphism in the 17th and 20th introns of the RB1 gene in Japanese and its application to genetic counseling in hereditary retinoblastoma.

AUTHOR: Ninomiya Shinsuke

AUTHOR ADDRESS: Dep. Pediatrics, Okayama Univ. Med. Sch., Okayama 700**
Japan

JOURNAL: Okayama Igakkai Zasshi 106 (1-2):p1-10 1994

ISSN: 0030-1558

DOCUMENT TYPE: Article

RECORD TYPE: Abstract

LANGUAGE: Japanese; Non-English

SUMMARY LANGUAGE: Japanese; English

ABSTRACT: Risk estimation for siblings or offspring is important in genetic counseling of patients with hereditary retinoblastoma. The RB1 gene spans approximately 200 kb in length, containing 27 exons. The use of polymorphic markers within the RB1 gene will eliminate the need of laborious specification of a mutation. The present study determined types and frequencies of **VNTR** polymorphisms of the 17th and 20th introns of the RB1 gene in 50 unrelated Japanese, using **PCR** amplification. In the 17th intron **VNTR**, there were 4 **alleles**, which ranged from 1400 bp to 1550 bp. The most common **allele** was 1400 bp with a frequency of 73%, and the heterozygosity rate was 46%. In the 20th intron **VNTR**, there were at least 9 **alleles**, which ranged from 192 bp to 240 bp. The **alleles** were more evenly distributed than those of the 17th intron **VNTR**, and the heterozygosity rate was 64%. These **VNTR** polymorphisms were successfully applied to the prediction of retinoblastoma and to the determination of parental origin of a chromosome deletion in 3 families with hereditary retinoblastoma. Analysis of **VNTR** polymorphisms within the RB1 gene proves to be practical and efficient for risk estimation in hereditary retinoblastoma.

1994

5/3,AB/149 (Item 25 from file: 5)
DIALOG(R)File 5:Biosis Previews(R)
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09230174 BIOSIS NO.: 199497238544

Allele frequency estimation of intragenic Bcl/I and XbaI RFLPs and extragenic St14 **VNTR** markers of the factor VIII gene and linkage analysis of 8 hemophilia A families.

AUTHOR: Kim Jang Seong(a); Oh Dahl Kyun; Kang Shin Hye; Moon Hong Mo

AUTHOR ADDRESS: (a)Mogam Biotechnology Res. Inst., 341 Pojung-ri,
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JOURNAL: Korean Journal of Genetics 15 (4):p295-306 1993

ISSN: 0254-5934

DOCUMENT TYPE: Article

RECORD TYPE: Abstract

LANGUAGE: Korean; Non-English

SUMMARY LANGUAGE: Korean; English

ABSTRACT: With the aid of **PCR**, we used two intragenic factor VIII gene RFLP markers (BclI-intron 18 and XbaI-intron 22) and one extragenic marker (St14 **VNTR**) to estimate **allele** frequencies of these markers from 100 Korean individuals and to analyze eight Korean hemophilia A families. **Allele** frequencies for the presence of the enzyme sites and heterozygosity expectations were 86% and 24% for

BclI-intron 18 RFLP marker, and 62% and 47% for XbaI-intron 22 RFLP marker, respectively. When BclI-intron 18 was homozygous, 31.5% of women was predicted to be heterozygous for Xba-intron 22. Number of repeats in St14 VNTR loci from Korean males differed from those previously reported for Caucasian males. 68% of Korean women was estimated to be heterozygous for St14 VNTR marker. Analysis of hemophilia A families with BclI-RFLP and St14 VNTR proved to be informative for six out of eight families tested.

1993

5/3,AB/150 (Item 26 from file: 5)
DIALOG(R)File 5:Biosis Previews(R)
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09131706 BIOSIS NO.: 199497140076

The use of PCR techniques for the VNTR allele
distribution analysis of 120 unrelated Russian individuals living in
Moscow.

AUTHOR: Chistyakov D A; Gavrilov D K; Ovchinnikov I V; Nosikov V V
AUTHOR ADDRESS: Res. Inst. Genet. Sel. Ind. Microorg., Moscow 113454**USA
JOURNAL: Molekulyarnaya Biologiya (Moscow) 27 (6):p1304-1314 1993
ISSN: 0026-8984
DOCUMENT TYPE: Article
RECORD TYPE: Abstract
LANGUAGE: Russian; Non-English
SUMMARY LANGUAGE: Russian; English

ABSTRACT: Allele frequencies of four VNTR regions (loci D1S80, D17S30, APOB and IGIIJ) were determined in 120 unrelated Russian individuals living in Moscow. The high level of length-polymorphism was discovered among alleles of these VNTRs. The genotype distribution of these hypervariable regions was established on the basis of experimental data. The comparative analysis showed the likeness between the allele distributions of these VNTRs among Russians and other groups of Caucasians living in Europe and North America.

1993

5/3,AB/151 (Item 27 from file: 5)
DIALOG(R)File 5:Biosis Previews(R)
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08935812 BIOSIS NO.: 199396087313

Limiting detection of an amplification signal for HLA-D region and
VNTR genes by phosphorus-32 PCR.

AUTHOR: McDaniel D Olga(a); Naftilan Janice; Barber W Henry
AUTHOR ADDRESS: (a)Dep. Surgery/Med., Univ. Ala. Birmingham, Birmingham, AL
35294**USA
JOURNAL: Biotechniques 15 (1):p140-142, 144-145 1993
ISSN: 0736-6205
DOCUMENT TYPE: Article
RECORD TYPE: Abstract
LANGUAGE: English

ABSTRACT: The limiting detection signal for identification of human genetic markers, such as HLA-D and VNTR genes, was determined using DNA isolated from a series of decreasing numbers of lymphocytes carrying the target marker in the polymerase chain reaction (PCR). The PCR procedure was assembled by incorporating 32P-labeled dCTP in the reaction mixture. Primers specific for detection of MHC Class II genes such as HLA-DR1, -DR2, -DRw52 and -DRw53 were utilized when cells were mismatched

by one DR type, and primers for the identification of the region of variable number of tandem repeats (VNTRs) were utilized where cells had the same DR types. The 32P-incorporated amplified DNA was analyzed by polyacrylamide gel electrophoresis followed by exposure to x-ray film. The sensitivity of the test varied for different allelic markers as evaluated by amplification of DNA from each set of a mixture of lymphocytes. The target HLA-DR markers were detectable in a cell ratio of as high as 1;100,000, whereas the VNTR markers were detectable at a 1:1000 cell ratio. The approach described here offers certain advantages: 1) increased sensitivity, 2) quantitative power, 3) reduced assay time, 4) simplified procedure and 5) less expense. This method provides valuable information for studies involving forensic specimens and marrow engraftment after allogeneic bone marrow transplantation (BMT) that require discrete representation of one allele relative to another in a heterozygous sample where limited quantities of target DNA are available.

1993

5/3,AB/152 (Item 28 from file: 5)
DIALOG(R)File 5:Biosis Previews(R)
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08472923 BIOSIS NO.: 199344022923
Heteroduplex analysis can increase the informativeness of VNTR polymorphisms amplified by PCR: Application to Stickler Syndrome.
AUTHOR: Wilkin D J; Cohn D H
AUTHOR ADDRESS: Cedars-Sinai Med. Cent., Los Angeles, Calif**
JOURNAL: American Journal of Human Genetics 51 (4 SUPPL.):pA205 1992
CONFERENCE/MEETING: 42nd Annual Meeting of the American Society of Human Genetics, San Francisco, California, USA, November 9-13, 1992. AM J HUM GENET
ISSN: 0002-9297
RECORD TYPE: Citation
LANGUAGE: English
1992

5/3,AB/153 (Item 29 from file: 5)
DIALOG(R)File 5:Biosis Previews(R)
(c) 2002 BIOSIS. All rts. reserv.

08355451 BIOSIS NO.: 000094095974
SOME VNTR LOCI TYPING FROM SINGLE HAIRS
AUTHOR: LEE J B
AUTHOR ADDRESS: DEP. FORENSIC MED., SEOUL NATL. UNIV. COLL. MED., SEOUL 110-460, KOREA.
JOURNAL: SEOUL J MED 32 (4). 1991. 209-215. 1991
FULL JOURNAL NAME: Seoul Journal of Medicine
CODEN: SJMEE
RECORD TYPE: Abstract
LANGUAGE: ENGLISH

ABSTRACT: Hairs were taken from 11 families (57 individuals) and peripheral blood was also drawn from 5 out of the 11 families (28 individuals). DNAs isolated from the hairs and peripheral bloods were amplified by polymerase chain reaction (PCR) with primers of the D1S80 locus, D17S30 locus, and the 3' hypervariable region of the apolipoprotein B gene. The fragment length polymorphism of amplified products (AMP-FLP) was analyzed by acrylamide gel electrophoresis followed by ethidium bromide staining. The recovery of DNA from a single hair was variable from person to person and even in an individual, 174 ng/hair on average, which was sufficient for analysis of the 3 genetic loci. The MAP-FLP of

the hairs was identical to that of the peripheral blood in all 3 genetic loci examined. The inheritance pattern of the 3 genetic loci was analyzed in amplified products of hair DNA, demonstrating exclusively Mendelian inheritance in all 11 families. Using hair DNAs of the 11 families, 15 **alleles** were detected in the D1S80 locus, 11 **alleles** in the D17S30 locus, and 9 **alleles** in the 3' hypervariable region of apolipoprotein B gene. Thus, analyses of genetic loci could be performed using hair DNA instead of peripheral blood DNA, when an appropriate amplification method is available and blood sampling is for some reason difficult.

1991

5/3,AB/154 (Item 30 from file: 5)
DIALOG(R)File 5:Biosis Previews(R)
(c) 2002 BIOSIS. All rts. reserv.

07750540 BIOSIS NO.: 000092064261

APPLICATION OF DNA AMPLIFICATION BY **PCR** IN THE STUDY OF HYPERVARIABLE
VNTR REGIONS FOR FORENSIC PURPOSES EXPERIENCE WITH APOB AND YNZ 22
SYSTEMS

AUTHOR: GIORGETTI R; TAGLIABRACCI A; CINGOLANI M; FERRARA S D
AUTHOR ADDRESS: IST. MED. LEGALE DELL'UNIV. ANCONA, SEIONE ANCONA, ITALY.
JOURNAL: BOLL SOC ITAL BIOL SPER 67 (1). 1991. 25-30. 1991
FULL JOURNAL NAME: Bollettino della Societa Italiana di Biologia
Sperimentale
CODEN: BSIBA
RECORD TYPE: Abstract
LANGUAGE: ITALIAN

ABSTRACT: The **PCR** method has been applied to amplify two
Variable-Number-Tandem-Repeat (**VNTR**) sequences. The high
polymorphism of these **VNTR** systems can be usefully applied in
medical legal fields such as paternity testing and individual
identification. The **VNTR** systems utilized were: ApoB and YNZ 22.
The study was conducted on a three-generation family of thirteen members,
whose relationship was previously established using conventional blood
systems. The results confirm the Mendelian inheritance of the
alleles found and the suitability of the **PCR** method for
forensic purposes.

1991

5/3,AB/155 (Item 31 from file: 5)
DIALOG(R)File 5:Biosis Previews(R)
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07498907 BIOSIS NO.: 000091072776

ANALYSIS OF THE **VNTR** LOCUS DIS80 BY THE **PCR** FOLLOWED BY
HIGH-RESOLUTION PAGE

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JOURNAL: AM J HUM GENET 48 (1). 1991. 137-144. 1991
FULL JOURNAL NAME: American Journal of Human Genetics
CODEN: AJHGA
RECORD TYPE: Abstract
LANGUAGE: ENGLISH

ABSTRACT: Allelic data for the D1S80 locus was obtained by using the
PCR and subsequent analysis with a high-resolution, horizontal PAGE
technique and silver staining. Compared with RFLP analysis of **VNTR**

loci by Southern blotting, the approach described in this paper offers certain advantages: (1) discrete **allele** resolution, (2) minimal measurement error, (3) correct genotyping of single-band **VNTR** patterns, (4) a nonisotopic assay, (5) a permanent record of the electrophoretic separation, and (6) reduced assay time. In a sample of 99 unrelated Caucasians, the D1S80 locus demonstrated a heterozygosity of 80.8% with 37 phenotypes and 16 **alleles**. The distribution of genotypes is in agreement with expected values according to the Hardy-Weinberg equilibrium. Furthermore, the observed number of **alleles** and the level of heterozygosity, obtained through the protocol described here, are congruent with each other in accordance with the expectation of a mutation-drift equilibrium model for a single, homogeneous random-mating population. Therefore, the analysis of D1S80 and similar **VNTR** loci by amplified fragment length polymorphism (AMP-FLP) may prove useful as models for population genetic issues for **VNTR** loci analyzed by RFLP typing via Southern blotting.

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L1: Entry 1 of 20

File: PGPB

May 9, 2002

PGPUB-DOCUMENT-NUMBER: 20020055628

PGPUB-FILING-TYPE: new

DOCUMENT-IDENTIFIER: US 20020055628 A1

TITLE: Multilocus repetitive DNA sequences for genotyping bacillus anthracis and related bacteria

PUBLICATION-DATE: May 9, 2002

INVENTOR-INFORMATION:

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Jackson, Paul J.	Los Alamos	NM	US	

US-CL-CURRENT: 536/23.7

Full	Title	Citation	Front	Review	Classification	Date	Reference	Sequences	Attachments	Claims	KWIC
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☐ 2. Document ID: US 20020012924 A1

L1: Entry 2 of 20

File: PGPB

Jan 31, 2002

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PGPUB-FILING-TYPE: new

DOCUMENT-IDENTIFIER: US 20020012924 A1

TITLE: Materials and methods for identifying and analyzing intermediate tandem repeat DNA markers

PUBLICATION-DATE: January 31, 2002

INVENTOR-INFORMATION:

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Bacher, Jeffery W.	Madison	WI	US	

US-CL-CURRENT: 435/6; 435/91.1, 536/24.3

Full	Title	Citation	Front	Review	Classification	Date	Reference	Sequences	Attachments	Claims	KWIC
Draw Desc	Image										